

The HCU Herald

HCU AWARENESS MONTH EDITION!

Featuring...



HCU Hero
Christa from Ohio



GO BLUE FOR HCU

October 2023



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



HCU HERO: CHRISTA FROM OHIO

My name is Christa, I'm 38 years old and live in central Ohio.

I was born prematurely on February 2nd, weighing just 3lbs 3 1/2 oz and measuring 19 inches in length, and from the beginning, my path was set on a unique trajectory. An out-of-range result on my newborn screening test prompted further investigation, leading to the discovery of homocystinuria through a concentrated urine test at 3 months old. My mother was able to breastfeed me initially and I later began my journey on a methionine-free formula with cystine, a regimen that proved unpleasant, yet essential. I required monthly appointments in Denver, Colorado for regulation of my homocysteine and methionine levels.



When I was 18 months old, my care transitioned to the Children's Hospital in Cincinnati and then to Ohio Children's Hospital in Columbus at age 12 through the present day. Throughout my childhood, my most notable symptom was digestive discomfort, likely attributed to the formula. The word "formula" was a word I'd shutter when hearing, as it represented the arduous battle that marked my daily routine - the confrontation with the prescribed formula. A relentless skirmish transpired after every breakfast and dinner, a true struggle, testing both my willpower and my parents' unwavering dedication to my health.

Imagine, if you will, facing a horrendous adversary in the form of a large glass of liquid that appeared innocuous yet concealed a bitter, sulphuric flavor with the texture of coarse sand water. This elixir, whose gritty texture would mockingly scrape against your teeth, demanded to be consumed, as it held the key to the necessary nutritional power my developing body craved to properly function. Yet, with each encounter, I found myself entangled in a battle of wills, desperately trying to evade its grasp, while my determined parents valiantly sought to ensure its ingestion.

To merely drink this concoction was an ordeal, for swift consumption risked triggering an involuntary gag reflex. The clump of grit within, like an ambush, would blast the back of my throat, threatening to undo all efforts. The very act of swallowing this formula became an intricate dance of caution and determination, a moment where victory was defined by distracting me enough immediately after swallowing it, to avert the reflex to expel what I had reluctantly consumed.

The consequences of this struggle extended beyond the immediate discomfort. Should I manage to retain the formula, a lingering unease would ensue, as my stomach churned for approximately 20 minutes post-consumption. The endeavor to maintain my health and manage my condition often resulted in an all-out battle of the ages every round. The formula became both a physical and emotional battleground, shaping my oppositional defiance to authority and my distaste for “healthy nutrition”, feeling that my parents’ need to micromanage my food choices to simply prevent whatever threatening adverse reaction scare tactics the doctors had warned them against was more of a ridiculous overprotective nuisance. I had never met

“
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”

another individual with homocystinuria, so began to feel it couldn't possibly be as serious as they had been dramatizing it to be, and it was simply a way they were trying to control me.

My family moved to Tokyo, Japan during my 8th grade through 10th grade school years and treatment for my homocystinuria was transferred to a specialist there. Honestly, treatment in Tokyo was so much more convenient! They individually packaged dosages of betaine to be easily kept in a lunch box and just tore open and dumped into whatever I had to drink at the time. It never was obvious to peers that I had anything different, as it just looked like I was using a water flavoring packet. So, I was more than happy to work it into my daily routine during meals and stash extra packages in my backpack, in case I was out later than expected. There I seemed to have so many more options for "normal meals" as rice was a staple for everyone, and fresh fruit & veggies were a normal staple as a side item. I simply appeared as a Vegan. I did still struggle with my formula there, though I no longer really fought to get it down. My levels were the best during that time period, which goes to show when options are available to allow children and teens to just blend in with daily norms and not be forced to stand out it not only improves their mental health, but also allows them to build a greater independence and ownership in maintaining positive control of their physical health.

When I returned back to the States in my 11th grade year, the independence and healthy routine that I had been thriving with came to a screeching halt when nothing was again convenient. I immediately felt disregarded by the medical and pharmaceutical world, now knowing there were much more accessible options elsewhere. I felt that because homocystinuria is more rare and not as lucrative of a money maker as other conditions, I was being disregarded and pretty much categorized as the dirt under the totem pole of worthwhile investments, left to nature's survival of the fittest; natural selection of only the strongest most viable will survive. I mean, we see how

quickly individuals can monitor their glucose or INR levels on the spot and are able to make immediate dietary adjustments, yet we instead must drive to the nearest lab, wait a couple of days for them to process the labs, then get our results back, and only then are finally sent diet or formula adjustment recommendation. Medication and treatment advancements have come so far for those areas because it's a great money maker! So then for us to get access to a life-altering treatment that could significantly improve quality of life we get shafted in supply because since the demand is low, the motivation to create quality products is too. Or, a product is made and the company decides they still want to make an equal profit as if it were to sell to a high-demand market, so they just charge the individual the total difference to make up for the amount they won't sell.

Despite dietary restrictions and watching my friends enjoy tempting treats, my resilience grew as I continued to push the line to access the options richer in flavor. It became my mission to figure out how to get flavors of the forbidden higher protein foods that my brain seemed to crave, and I happily traded peers at lunch for nibbles of their more enticing entrees. I navigated school by trying to mask my continued disappointment and frustration with having to just barely skim by in math, test-taking, and completing multi-step assignments (due to significant executive function deficits) by engaging in avoidance tactics, keeping busy with the social scene of high school, and pretending to not let it bother me. I skirted under the radar of struggling behind my peers until it was advised to my parents that if I wanted to make it through college, I should have a cognitive neuropsychological evaluation for possible learning disabilities. I underwent an evaluation, which confirmed a diagnosis of ADHD and a math learning disability. This formally opened the doors to access accommodations for extended time on tests, provided notes for classes so I could focus my attention on soaking up the lectures versus trying to split my attention with the notetaking, and allowed me to access free additional tutoring for math classes. I additionally started on ADHD

medication which was a game-changer in my ability to focus in class, manage my class requirements, and complete multi-step projects.

College life was a struggle to manage my intake of medication, as I had a typical college student's routine (or lack thereof!) I never wanted to take all my stuff that I'd need to take my formula to the food court, as it took up too much time to put together (I still lacked the executive function skills and strategies to manage my time to allow extra allotted time for my formula needs to ensure I got it in every day). My newfound freedom in college of not having to battle my parents to get down my formula and to finally have control of how I wanted to manage it won more often than that voice in the back of my head that knew I really should be getting it in. I persuaded myself that still following the low protein diet (especially with the big ticket items of no meat or high protein foods) was sufficient, and if I wanted a sprinkle of cheese for flavor or one piece of sausage on my pizza for flavor was fine because I'd just balance it out with the next meal by having a salad. My experimental dance with the balancing of meals and lack of consumption of the daily formula was a roll of the dice, and no physical symptoms were noticeable during that time. My homocysteine and methionine levels, however, were much more noticeable and elevated.

“My experimental dance with the balancing of meals and lack of consumption of the daily formula was a roll of the dice, and no physical symptoms were noticeable during that time. My homocysteine and methionine levels, however, were much more noticeable and elevated.”

As I matured into a young adult, my homocystinuria was always on the back of my mind when dating, as at the time I had been told I'd be too high of a risk to have kids and was aware that the science at the time had indicated the average life-span of individuals with my condition was only to the age of 30-something.

I didn't necessarily feel like I was the ideal "catch". But I knew that part of my non-negotiables in searching for Mr. Right had to include someone open to adoption, and that would understand the potential risks involved; meaning that I may not be the person he'd get lucky to "grow old with". My husband and I met and started dating in our senior year of high school! 5 years later, after finally marrying my Mr Right, I made sure one of the first things my husband understood was how to spot the signs of a stroke, as I knew that this was one of the serious potential risks I faced as someone with HCU. I made sure to review our plan often of what he was to do if he saw those signs.



In July 2009, we adopted a 6-year-old boy with special needs, who is now 21. As the years progressed and we battled with my son's special needs, I continuously worried about how my husband would manage the complexities of our son's needs by himself, if something were to happen to me.

In 2015, when my son was 12 and I was 30, that fear was tested as close to reality as I could have ever predicted. I woke up on the morning of December 26th, the day after Christmas, and jumped in the shower. I had a sharp charlie horse in the bottom of my left breast that didn't ease up. "WEIRD", I thought. "Maybe I slept on it wrong.", and I continued my shower. Then I had a sudden onset of severe nausea and became sick in the shower, followed by dizziness so overwhelming that I had to sit on the floor of the bathtub. At that point, I knew that time was of the essence. I summoned my husband to get me to the car and take me to the emergency room.



At first, the medical team waived it off as a panic attack induced by holiday stress, but the doctor and his team quickly went on high alert after my EKG showed a heart attack. I coded immediately afterward, and several more times again. They informed my husband that he most likely wasn't going to get to take me home, as they didn't think I'd make it before they could get me transferred to a different hospital and into surgery. It turns out I had a blood clot in the lower left ventricle of the heart, or what they call "the widowmaker" (due to the severity and often difficult location it is to get in and repair). This was a result of a high homocysteine level.

The blood thinners they had put me on ended up becoming subtherapeutic, and in November 2021, caused a massive stroke due to two blockages; one in my carotid artery and another at the cross of my basal ganglia and prefrontal cortex. However, thanks to the preparation, my husband quickly identified the

signs of the stroke. I had awoken in the middle of the night to let the dog out and had been stumbling around, had impaired speech, and my smile had gone completely crooked. The emergency squad was there within 15 minutes. We were told that my husband's fast response was what got me the assistance I needed within the "golden hour" as they say, which allowed them to rush me to surgery and remove the clots in enough time to allow 90% of my damage from the stroke (as of today) to be reversed.

Today, we are officially in the process of trying to get pregnant for round #2 of our parenting journey! We've had to assemble a VIP team of medical professionals to craft a meticulous plan of preparation and treatment during & after the entire process. While this is an exciting time, I know that it will not be an easy journey with having to stay on blood thinners throughout the pregnancy and potentially some of my heart medications. Therefore, I am forced to give over my worries to prayer, that this medication won't cause harm to a future baby.

My personal journey with homocystinuria, navigating my own learning challenges, and supporting my son through his own learning struggles, combined with 13 years of experience as an Intervention Specialist and 6 years in the Educational Therapy field, have shaped me into the professional I am today. These experiences empowered me to establish [RootED Learning & Development Center](#)

in July 2020. At RootED Learning, I offer personalized 1:1 Educational Therapy, ADHD Coaching, Executive Functioning Coaching, College & Career Readiness guidance, and advocacy for IEP/504 plans. My services are available to individuals ages 8 to adults of all ages, both locally in central Ohio and virtually across the U.S. & Canada.



To read Christa's story on our website, visit <https://hcunetworkamerica.org/christa/>

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



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POWDER

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VEGGIE DUMPLINGS



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Eat Well. Live Well.



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NEW RESOURCE

Let's help you Transition to Adulthood!

We're adding more resources to supplement our *Transition to Adulthood* milestones guide!

Insurance



I know what the terms 'deductible', 'co-pay' and -out-of-pocket' mean.

[CLICK HERE](#)

to download this resource

I know what the terms "deductible", "co-pay" and "out-of-pocket" mean..



Premium

The amount you must pay on a monthly basis for your insurance plan.

Deductible	Co-Pay	Coinsurance
<p>The total amount you pay for healthcare services before your insurance starts covering the costs.</p> 	<p>A fixed dollar amount that you pay out of pocket for covered services.</p> <p>Your co-pay may differ depending upon the type of care you seek (ex. primary care, specialist, emergency care).</p> 	<p>A percentage of a covered service that you pay after you've already paid your deductible.</p> 
<p>Out of Pocket Maximum</p> <p>The maximum amount that you can pay toward your covered services in any given year. This includes deductibles, copayments, and coinsurance.</p>		

Let's see this in action

You have bloodwork that costs \$400. Let's say you've already met your deductible. With coinsurance of 20%, how much would you be paying out of pocket?

 Cost of visit \$400	×	 Your coinsurance 20%	=	 Your payment \$80
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Tip! Download your insurance companies app to track the progress of your deductible!



[Click here](#) to access our *Transition to Adulthood* guide & additional resources!

WEAR YOUR AWARENESS

October is HCU Awareness Month!
Get your Gear NOW!

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



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

GO BLUE FOR HCU

October is HCU Awareness Month

HCU Network America — Hcunetworkamerica.org — info@hcunetworkamerica.org

HCU AWARENESS MONTH - SOCIAL MEDIA PLAN!

- ♥ One of the best ways to reach a wide audience to spread awareness for HCU is through social media!
- ♥ We'll be asking you to participate in different ways throughout the month (see below). Don't forget to use the #hashtags & tag us in your posts!!!

October is HCU Awareness Month!

October 2-6: Intro to the Homocystinurias

October 9-13: Focus on Classical HCU

October 16-20: Focus on Cobalamin Defects

October 23-27: Focus on Severe MTHFR

#Homocystinuria

#HCUAwareness2023

#HOPECONNECTSUS

#GOBLUEFORHCU

How can you spread awareness?

MONDAY



Re-share
our
infographic
for the
week

TUESDAY



Share the
incidence
of your
condition

WEDNESDAY



Create an
image with
you and
arrows
pointing
to your
symptoms

THURSDAY



Make a
video
sharing
what your
treatment
looks like

FRIDAY



Share
your
patient
story

Follow us!

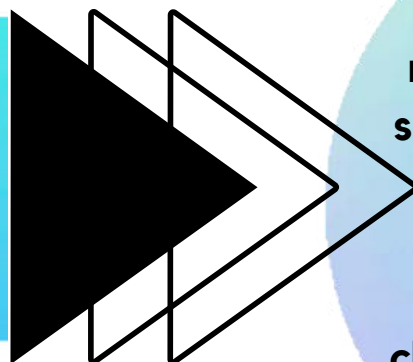


It's HCU Awareness Month – **LET'S TALK FUNDRAISER SETUP!**



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

One way is by
hosting a
fundraiser!



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

Not sure how to get started?

Let our Fundraising committee help!

Email
info@HCUnetworkamerica.org

Click [here](#) for a list of additional HCU Awareness and Fundraising Event Ideas!

HCU Awareness Month



Activity List



We know this is a rather long list, but we love your participation as it helps raise awareness for our small but mighty community. Please try to do as many as you can. If you do one each day, you'd complete the list!

- Change your social media picture to the HCU Awareness Ribbon
- Start a HCU fundraiser
- Share an infographic about HCU
- Share a patient story
- Share your diagnosis story
- Challenge your friends to the same amount of protein and three normal protein shakes a day #ToastToHCU
- Share a pic of an item that has the same amount of protein you can have
- Share your daily diet record -completed
- Share a low-protein meme
- Share your favorite low protein recipe! Bonus if you cook it and share a pic
- Dining out, low protein style. Where do you like to eat?
- Share a pic of what your grocery store haul looks like
- Real cost of HCU: Grocery Cost Comparison #Medical Nutrition Equity Act. or share some patients with HCU require injectable B12. B12 on average is \$300-400 a month and most insurance companies don't cover it!
- Share a picture or video capturing all the medication you take (this includes formula for those who need it).
- Share a picture of your first pair of glasses, or a device that helps you navigate or communicate due to lack of vision
- Share something you wish people understood about HCU
- #HaikuforHCU—Write and share a Haiku describing life with HCU
- Wear jeans for your rare genes #ItsInOurGenes
- Wear your HCU Shirt and share a pic online—#HopeConnectsUs
- #GoBlueforHCU
- #HCUAwareness post in a public place
- Share with a stranger what HCU is and why it's important to you
- #Create4Cure—Create a work of art that brings awareness for HCU— can be a song, dance, a painting—get creative!
- #High5forHCU—List 5 ways HCU makes you a stronger, better person
- All states test for classical HCU, but many are still missed
- Share a picture of you and a HCU buddy! Or tag a friend who is of great support
- #FacesofHCU—Share a picture of you saying, I am one of the 1 in 200,000 people with HCU
- #Hope4HCU—Share 4 things that give you hope and encouragement
- Share the HCU timeline—if you know other facts, let us know!
- Cutting Edge of HCU: Share about a therapy that is in the works!

To find additional information and resources, visit:
<https://hcunetworkamerica.org/hcu-awareness-month/>

OUR MATCHING GIFT IS BACK!!!

That's right, you heard us right! Another anonymous donor has joined (we went from 2 to 3!). That means that **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. Set up a Facebook or Instagram Fundraiser, Give Lively, GoFundMe, or host your own alternative fundraising event and invite your family and friends to participate! Alternatively, they can donate directly to HCU Network America. **Anyone who creates a fundraiser and raises over \$100, will receive a HCU Awareness car magnet!**

Have an idea for a fundraiser, but not sure how to get started? Let our fundraising committee help you get started Email info@hcunetworkamerica.org and we will connect you!

Matching Gift Challenge

Three generous donors have pledged to match EVERY gift up to \$25k of donations received until December 31!

\$25,000



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?



Meet our Fabulous Fundraisers!

With **7,808** total miles logged, our Race Participants & Fundraisers raised an astounding **\$45,463** for HCU research grants!



Amy's Army
\$1,305



Masen's Mad Dawgs
\$1,275



Team Anniston
\$1,025



Brooke's Blazers
\$2,175



Mighty Marchese's
\$75



Team Eton
\$4,115



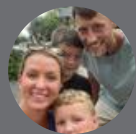
Cakes for Carson
\$4,540



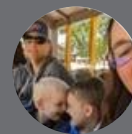
Miles for Marley
\$2,150



Team Hawkins/PwC
\$1,969



Carter Crew 4 HCU
\$75



Race with C & G
\$75



Team Recordati
\$1,130



Ellie's Entourage
\$2,425



Recordati - Sylvia's
Supporters
\$720



Team Will for HCU
\$2,150



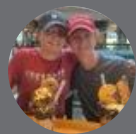
Grayson's Gang
\$1,363



Renna's Rare Runners
\$680



The Bartke Ruff Ruffs
\$845



"Hunt" for research
\$16,146



Synlogic
\$430



The Rare Runners
\$60

Grants will be awarded. or grant applications will open...(Info here??)

FEATURED FUNDRAISER



"Hunt" for Research



The Hunt family's fundraiser raised an astounding **\$16,146** for Cobalamin G research!



Carson biked a total of **116 miles!**

Cobalamin G is a rare form of Homocystinuria. With only 50 identified cases worldwide in the literature, the need for research is critical.



Ready to feel inspired? Check out Carson's story on our Youtube channel!



<https://bit.ly/3fmCXzR>

HCU HEROES

RACE FOR RESEARCH



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



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[Learn more about Recordati Rare Diseases](#)

Cheer Section Sponsors



[Learn more about Cosette Pharmaceuticals](#)



[Learn more about Synlogic](#)

Whether you reach for Vitaflo™ HCU express™ plus for its low volume or mixing flexibility, it allows you to "express yourself."
 Mix with 3 fl oz of water or customize it to match your style and taste at any given moment OR any given time OR any time you want!



Make it. Shake it. Take it.

- ✓ Suitable from 3 years of age
- ✓ Pre-measured powdered formulas containing 15 g or 20 g protein equivalents (PE) per packet
- ✓ Mix with approximately 3 fl oz water or other permitted beverages to a low volume drink
- ✓ Unflavored

Check out our new **express™ plus patient guide:**



FEATURED RECIPE

Lemon Sorbet

- 1 packet **express plus20** unflavored
- 1 cup permitted lemon sorbet (slightly melted)

Place the ingredients into a blender and blend until smooth.

Nutrition Info

Calories 320 | PE20 g | Intact Protein 0.2 g

For our collection of low protein recipes, check out the recipes section on our website VitafloUSA.com/recipes

For more information about Homocystinuria (HCU) products and to request a sample, visit VitafloUSA.com

FOR USE UNDER MEDICAL SUPERVISION

Be sure to check with your healthcare professional before making changes to your diet.
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For assistance accessing Vitaflo products



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!



UPCOMING EVENTS

Our EL-PFDD, or Externally Led Patient-Focused Drug Development Meeting, with the FDA is happening this month!

Have questions?

To learn more about the purpose & format of an EL-PFDD meeting, as well as how you can participate, watch the replay of our Community Prep Webinar [here!](#)

CLASSICAL HCU EL-PFDD

October 27, 2023, at 10 am to 3:00 pm ET



KEYNOTE SPEAKERS



PANELS



MODERATED DISCUSSION




 INTERACTIVE LIVE POLLING | TELEPHONE CALL-INS 

Your attendance is critical! Click [here](#) to register to attend!


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.



WHAT NOW?
CLASSICAL HCU EL-PFDD
COMMUNITY DEBRIEF
REGROUP, REFOCUS, EXPLORE
November 05, 2023, 4 pm ET



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!

2024 Family Conference



Registration will open on **October 6th!**



June 29-30th 2024
Aurora, Colorado



- Free attendance for families!
- Low protein & regular menus provided!
- Kids & Teens programs!
- Fantastic opportunity to meet other families living with HCU!
- Informative sessions & workshops!
- Learn about the latest in research for HCU!
- Vendor freebies!

SPECIAL EDITION - SSIEM IN JERUSALEM!



Last year 20 patients & caregivers affected by Classical HCU took part in market research interviews. The data and themes from the conversations was then analyzed to understand the impacts, signs and symptoms, as well as desired changes for a potential new treatment.



This data was formatted into a poster presentation, which our Executive Director, Danae' Bartke, shared at the SSIEM (Society for the Study of Inborn Errors of Metabolism) Annual Symposium in Jerusalem last month! In addition, Danae joined a panel of speakers on the *Psychosocial impact of living with an IEM*, and also presented in the main session on "Understanding the Burden of Classical homocystinuria from the patient's perspective: a qualitative study", which was based on the poster developed from the market research findings.

You can view the poster [here](#):

We'd like to thank the patients and the caregivers who took part in these interviews. ***You are helping make a difference in the lives of those with HCU!***

Bone, Cartilage and Connective Tissue Survey

"...sometimes I feel like a Great Dane, like my body is stretched out, longer than it should be. I have had surgery on my left ankle, right knee (two surgeries), and right shoulder, and now am dealing with severe pain as a result of lumbar scoliosis. I sometimes think my body shouldn't feel like this at 45. "

- Andrew, Classical Homocystinuria patient



[Complete the Survey](#)

homocystinuria.rare-x.org



Acappella

NOW ENROLLING

ACAPPELLA Study on Classical Homocystinuria

Travere Therapeutics is enrolling children and adults with classical homocystinuria (HCU) in a ACAPPELLA Study. The goal is to learn more about classical HCU and the course of the disease. Information gained from this study may help to improve understanding of HCU and help other patients, families, healthcare providers, and researchers to design new clinical research studies and therapies. No investigational medicine will be given to participants.

Approximately 150 participants will take part at sites in the US, Europe, and other countries around the world. The study will include three key stages (screening, enrollment, and observational follow-up) and will last approximately 6.5 years.

You (or your child) may be eligible to participate in the ACAPPELLA Study if you:

- Have been diagnosed with HCU
- Are 1–65 years of age

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

For additional information about the ACAPPELLA Study, please go to:
<https://www.clinicaltrials.gov/ct2/show/NCT02998710>

You may be able to receive payment for time and travel when you participate in this study. Talk with your doctor and family members about joining the ACAPPELLA Study. Sites are open and currently enrolling participants.

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

If you have any questions, please email:

medinfo@travere.com

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



*Restrictions apply

MA-PE-22-0004. March 2023



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Request a Sample
3-Pack Today



SCAN TO REQUEST
A SAMPLE

WHERE WILL YOU DRINK YOUR
HCU LOPHLEX[®] LQ THIS FALL?

NutriciaMetabolics.com

*Contains 44.1% juice.

Brought to you by Nutricia North America.

HCU Lophlex LQ is a medical food for the dietary management of Homocystinuria for individuals over 4 years of age. Must be used under medical supervision.

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NUTRICIA



SCAN TO
STAY CONNECTED

Through our Nutricia Connect program, we provide families like yours with support at various stages of life's journey



LOW PROTEIN
RECIPES



PATIENT
EDUCATION



DIET MANAGEMENT
TIPS



PATIENT
CONNECTIONS



FORMULA COVERAGE
SUPPORT

Sign-Up Today at
NutriciaMetabolics.com/Nutricia-Connect

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NUTRICIA

Now Available!

Betaine Anhydrous for Oral Solution 180 gm

An AB rated Generic version of Cystadane®
(betaine anhydrous for oral solution) with full
patient support services you might expect from a Brand†



Copay support

Eton Cares can help eligible, commercially insured patients get their medication for **as little as \$0 per month***

*Restrictions, limitations, and/or eligibility requirements may apply. For patients who are not eligible for copay support or who need additional financial assistance, Eton Cares can help connect you with alternative forms of medication coverage or provide referrals to other possible sources of funding.



Financial support

Patients who do not have insurance and meet certain financial requirements may be eligible for additional financial support from our **Patient Assistance Program***

Eton Cares can provide copay and financial support.



**Have your doctor complete the
referral form to prescribe and enroll.**

Click Here

IMPORTANT SAFETY INFORMATION

Warnings and Precautions

Hypermethioninemia in Patients with CBS Deficiency: Betaine Anhydrous may worsen high methionine blood levels and accumulation of excess fluid in the brain has been reported. If you have been told you have CBS deficiency, your doctor will be monitoring your methionine blood levels to see if changes in your diet and dosage are necessary.

Adverse Reactions

Most common side effects were nausea and gastrointestinal distress, based on a survey of doctors.

To report a suspected adverse event related to Betaine Anhydrous, contact Eton Pharmaceuticals, Inc. at 1-855-224-0233 or the U.S. Food and Drug Administration (FDA) at <http://www.fda.gov/MedWatch> or call 1-800-FDA-1088.

INDICATIONS AND USAGE

Betaine anhydrous for oral solution is indicated in children and adults for the treatment of homocystinuria to decrease high homocysteine blood levels. Homocystinuria is a rare genetic disorder in which there is an abnormal accumulation of the amino acid homocysteine in the blood and urine. The following are considered to be homocystinuria disorders:

- Cystathionine beta-synthase (CBS) deficiency
- 5,10-methylenetetrahydrofolate reductase (MTHFR) deficiency
- Cobalamin cofactor metabolism (cbl) defect

eTon PHARMACEUTICALS

Please see enclosed Full Prescribing Information for more information.

†Cystadane is a registered trademark of Recordati Orphan Drugs SAS, not affiliated with Eton Pharmaceuticals.

1402-v2

Chiles Rellenos



Makes 6 | 2.55 grams protein per Chile Relleno

Ingredients:

- 6 Poblano peppers
- 1 cup Mixquick (you may be able to sub eggz or CFL Baking mix)
- 1 cup Dairy-free heavy cream
- 1 box Ener_G Foods bread crumbs
- 2 containers cheese wizard (Primal kitchen plant-based cheese dip may work also.)
- 20 oz salsa
- 1 can diced chilies
- Cooking oil, enough to submerge the chiles rellenos

Optional:

Taste connections bacon bits to taste (crumbled hooray bacon can be subbed)

Directions:

1. Grill the poblano peppers 5 minutes on each side till they are blackened, use tongs to flip. Transfer to a zip lock bag to sweat.
2. Once cooled down for a few minutes, peel the outer layer of skin off.
3. Mix together MixQuick and heavy cream until you get a pancake consistency. This can be done while the peppers are roasting.
4. In a small sauce pot mix together cheese wizard, salsa, diced chilis, and bacon bits if using.
5. The peppers normally will have a hole in them; use a small spoon (the smaller the better) to stuff the cheese sauce into the peppers, being careful not to tear them any further.
6. Close the openings with toothpicks as much as possible, and coat with MixQuick mixture and bread crumbs
7. Pour oil into a frying pan & wait for it to boil. put the Chiles Rellenos into the frying pan & cook for about 5 minutes on each side until they reach a golden-brown color.

This recipe is courtesy of our community's own Joanna Ball!

Click [here](#) to see this recipe made step-by-step, and visit Joanna's YouTube channel for more delicious Low Protein recipes!



THIS WEEK'S MENU

M

Breakfast: Breakfast Sandwich & Hashbrowns
Lunch: Veggie Wraps & Pretzels
Dinner: English Muffin Pizzas & Side Salad

T

Breakfast: Fruit, Yogurt & Granola Parfait
Lunch: Veggie Nuggets w/ Veggies & Ranch & Pretzels
Dinner: Spaghetti & Veggie Meatballs

W

Breakfast: Bagel & Cream Cheese w/fruit
Lunch: Grilled Cheese Sandwich w/tomato soup
Dinner: Asian Rolls & Rice

T

Breakfast: Waffles & **Enrich:** Mac n Cheese & Steamed Veggie Medley.
Dinner: Jackfruit tacos

F

Breakfast: Avocado Toast
Lunch: Burger & Prezel Sticks
Dinner: Balsamic Pasta Dish w/Foccacia Stick

Each day has meals for <10 grams (g) of protein/day, 20-30 g. of protein/day, and 30-40 g. of protein/day.

Shopping List

Click each day to view the week long menu!

Disclaimer: This meal plan is intended to be a foundation or guide to what meals could look like on a low protein diet. It does not take into account individual caloric, protein and formula requirements, which are all patient-specific. Please consult with your metabolic geneticist and dietitian prior to making any significant dietary changes or following any meal plans of which you are unsure.

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 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.

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.

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.



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>

**FOLLOW
US**

