The HCU Herald

2024

Featuring...

HCU Hero
Janet from California

January 2024

HCU NETWORK AMERICA

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

HCU HERO: JANET FROM CALIFORNIA

My name is Janet and I live in central California. I am thirty-one years old and live with classical Homocystinuria.

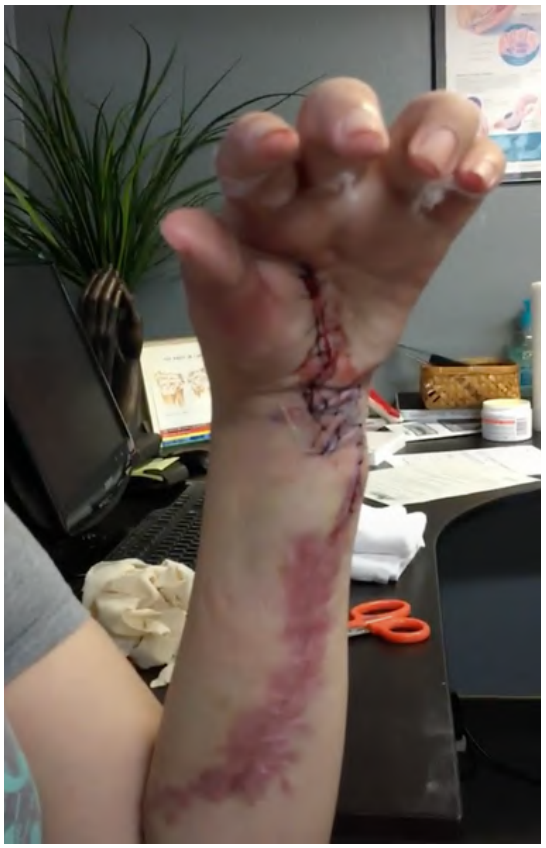
I want to take you back to the year 2002. I was ten years old, sitting with my parents in a tiny room at the Children's Hospital. Two doctors came into the room, who later turned out to be my dietician and genetics doctor. Hearing the word "Homocystinuria" for the first time, sounded like a voodoo spell. My parents looked just as confused as I did and had a million questions, like "Is it a terminal disease?" and "Will I be able to live a full life?" After getting some answers from the doctors, my parents seemed more at ease but still worried. I had no clue what was going on. I couldn't even wrap my head around what was being said. I just knew that my life was going to completely change.

As soon as we got home my parents tried to make as many changes to my diet as they could, and I fought them every single step of the way. I couldn't understand why I was the only one in the family that had to change the way I ate. Remember, I was just ten years old with NO understanding of what Homocystinuria was. To me it was just a word that branded me as an outsider. I felt like I was put on display for not only my family, but my classmates as well. In the cafeteria, other kids would look at my lunch and would ask me why I was eating "that" instead of what they were having, and I never could explain it because I myself did not know why I couldn't have a "normal" lunch.

HCU HERO: JANET FROM CALIFORNIA

I didn't know what was going on within my body. I wanted nothing more than to have the foods that I had always eaten. The embarrassment was too much for me and I was tired of being looked at differently because of my food. I felt like I was being punished without knowing why. I was struggling so hard to stay within the guidelines of my diet. My levels were not going down and at every doctor's visit, I would get scolded.

I vividly remember my doctor telling me that by the age of 5, a child develops most of their taste buds. I was ten when I was diagnosed. She said I was pretty much set up for failure. Had I been diagnosed earlier, I would not have struggled as much as I did. I still struggle heavily with the diet. The low protein diet is incredibly stressful, and difficult to maintain.



I recently have tried to regain control over my diet, but I ended up becoming even more anxious and scared to eat. I lost ten pounds in a month in the process and felt miserable and isolated. I obsessed over every morsel I put into my mouth. The diet has consumed me entirely; having to constantly count my protein intake and check the nutritional facts on every box of food. In any other situation where you're obsessing over nutrition labels and terrified of putting food in your mouth, they'd consider it an eating disorder. But somehow, this is the standard that we are prescribed, and it's considered healthy. At the end of the month, I realized I couldn't do it anymore. I wanted to be happy even if that cost me. I ended up paying the price: I developed a deep vein thrombosis in my left hand, and as a result, I've lost 60% of my arm function.

Being a first-generation Mexican-American and trying to navigate this disease took an extreme toll on me. Since the diagnosis was so rare, many doctors did not know how to explain it and there were even less people who knew how to interpret my disease in my parents' native tongue. So, there I was, a scared ten-year-old girl, having to translate my disease, that I knew nothing about, to my parents.

In a Hispanic household, food means love, family and togetherness. If you don't eat what is prepared, it can be taken as a sign of disrespect to whoever made the food. For Latinos, food and social gatherings are a core part of our culture. So, when I stopped eating due to my diet, everyone was concerned and scared for my health. I felt like an outsider in my own family. While everyone was enjoying their bistec con arroz, I was stuck eating salads.

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I long for the day when I'm not consuming 33 plus pills a day, but my biggest wish is to one day have a better treatment for HCU; one that would allow me to eat a less-restrictive diet, so that I can stop seeing food as the enemy, and truly feel a part of my family's celebrations again.

Click here to see Janet's video from our EL-PFDD meeting!

To read Janet's story on our website, visit:
https://hcunetworkamerica.org/janet/

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

Mississippi Jackfruit Roast

***Makes about 8 servings* | 1 serving = 5 oz | 1.7 grams protein per serving**

Ingredients:

- 140 g Jackfruit, canned, drained, seeds removed
- 340 g Raw Potatoes (1 pound), roughly 2 medium size potatoes
- 454 g Raw Baby Carrots, 1 pound bag
- 1 1/2 c Vegetable Broth
- 1 unit(s) Ranch Dips Mix, dry, 1 oz packet
- 1 unit(s) Brown Gravy Mix, dry
- 2 tsp White Wine Vinegar or Apple Cider Vinegar

Directions:

1. Main prep: Rinse and dry jackfruit and remove seeds. Wash potatoes and peel skins. Cut into medium chunks.

- Instant pot: Place the broth into the instant pot, followed by all the listed ingredients. Lightly stir to combine. Place the lid of instant pot on and turn valve to seal position. Press manual or pressure cook button and set time to 8 minutes. Once the instant pot finishes cooking allow to natural release for 5 minutes then release the rest of the pressure. Potatoes and carrots should be fork tender. If not, return the lid and valve to seal and pressure cook for another 2 minutes. Immediately remove pressure. Add salt and pepper to taste. Serve hot.

- Stove top: Place all ingredients in a large pot. Lightly stir to combine. Place over medium high heat to bring to a slow boil. Reduce heat to medium and cover. Allow to cook until the potatoes and carrots are fork tender. This may take thirty minutes. Add salt and pepper to taste and serve.

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*!

Scholarship opportunity!

flok The Guthrie-Koch Scholarship

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

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

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

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

The application deadline is March 15, 2024.

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!

Upcoming Events

HCU COMMUNITY TOWNHALL MEETING

Sunday, January 7, 2024 | 4 pm ET

Come see what we have in store!

TOPIC:

2024 Plans and Restructuring

SPEAKERS:

Danae' Bartke
Liz Carter
Dana Hunt
Brittany Parke
Grace Talbert

https://bit.ly/HCUTownHall

Raising Joy

Double Good

Give Good. Get Good. That's Double Good!

When you buy Double Good's award-winning, ultra-premium popcorn, 50% of your purchase goes to supporting our cause and you receive a delicious reward!

01.15.2024–
01.19.2024

THANK YOU!

Link to shop coming soon!

HCU NETWORK AMERICA

PANDA EXPRESS CHINESE KITCHEN®

+

HCU NETWORK AMERICA

When?

Friday, February 9, 2024

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

Where?

Panda Express locations nationwide

www.pandaexpress.com

How?

Online orders only.

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

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

GET READY....SUPER BOWL SQUARES ARE COMING!!!!

FOOTBALL SQUARES FUNDRAISER

FEBRUARY 11 | 6:25 PM ET

$10 PER SQUARE

4 CHANCES TO WIN!

VENMO: @TOM-HAWKINS-1

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

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

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

RARE DISEASE WEEK ON CAPITOL HILL

Registration opens January 3, 2024!

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

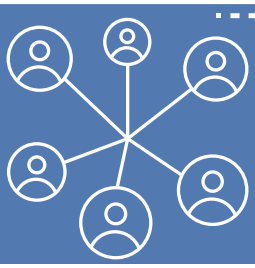
Hosted by the Rare Disease Legislative Advocates (a program of the EveryLife Foundation for Rare Diseases), this multi-day event brings together rare disease advocates from across the country to make their voices heard by their Members of Congress. Participants are educated on policy proposals impacting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

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

LET US KNOW!

Planning to attend Rare Disease Day events?

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



RARE DISEASE DAY IS COMING

MARK YOUR CALENDARS...

...for Rare Disease Day 2024!

Rare Disease Day @ NIH –
attend in person or virtually!

NIH main campus (Natcher Conference Center) and virtually on Thursday, Feb. 29, 2024, from 9 a.m. to 5 p.m. EST.

Click here for more information & to register to attend!

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

Grab your gear!

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

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

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!

Spotlight on Oral Health!

Last year there was a lot of chatter about dental health in the HCU community. In response, Dr. Kimberly Chapman issued the challenge last November to the HCU Community to enroll in RARE-X, take the Head to Toe Survey, and complete the Oral Health Survey. We utilized the data you contributed, and here are the findings

1 All types of HCU

Homocystinuria patients, regardless of type do not have an increased risk for gingivitis, but long-term risk for gum diseases

2 Cobalamin and MTHFR

Homocystinuria due to a cobalamin disorder or severe MTHFR often have some tooth structure differences.

3 Classical HCU

Reaffirms that classical HCU patients are more likely to have malalignment or malocclusion.

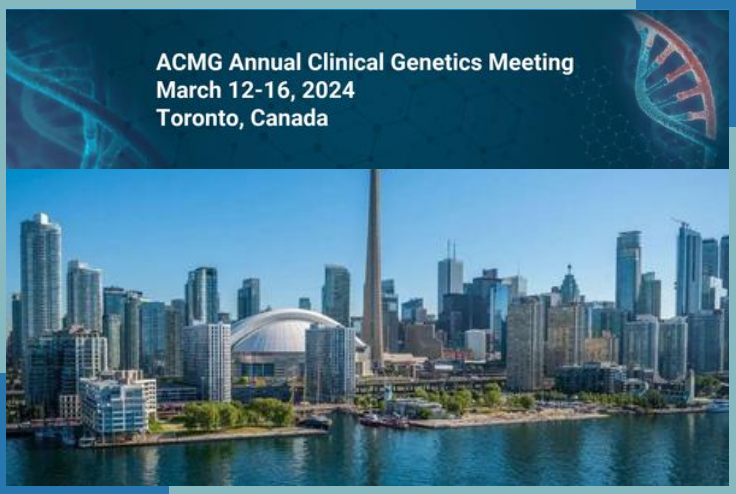
Read the full publication

https://doi.org/10.1016/j.ymgmr.2023.100999

BE COUNTED!

Your voice belongs in HCU Research

Your experiences can unlock a deeper understanding of the homocystinurias and accelerate new therapies. Regardless of where you are at in the globe, with Rare-X you can participate in research and it only takes a few minutes!



Help Dr. Chapman with her ACMG poster, take the Head to Toe Survey!

1. **Enroll or update your dashboard at https://homocystinuria.rare-x.org/**
   (5 minutes)

2. **Take the Head to Toe Survey**
   (15 minutes)

3. **Complete the other surveys in your dashboard by January 31!**

CANPCU+ CANPKU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

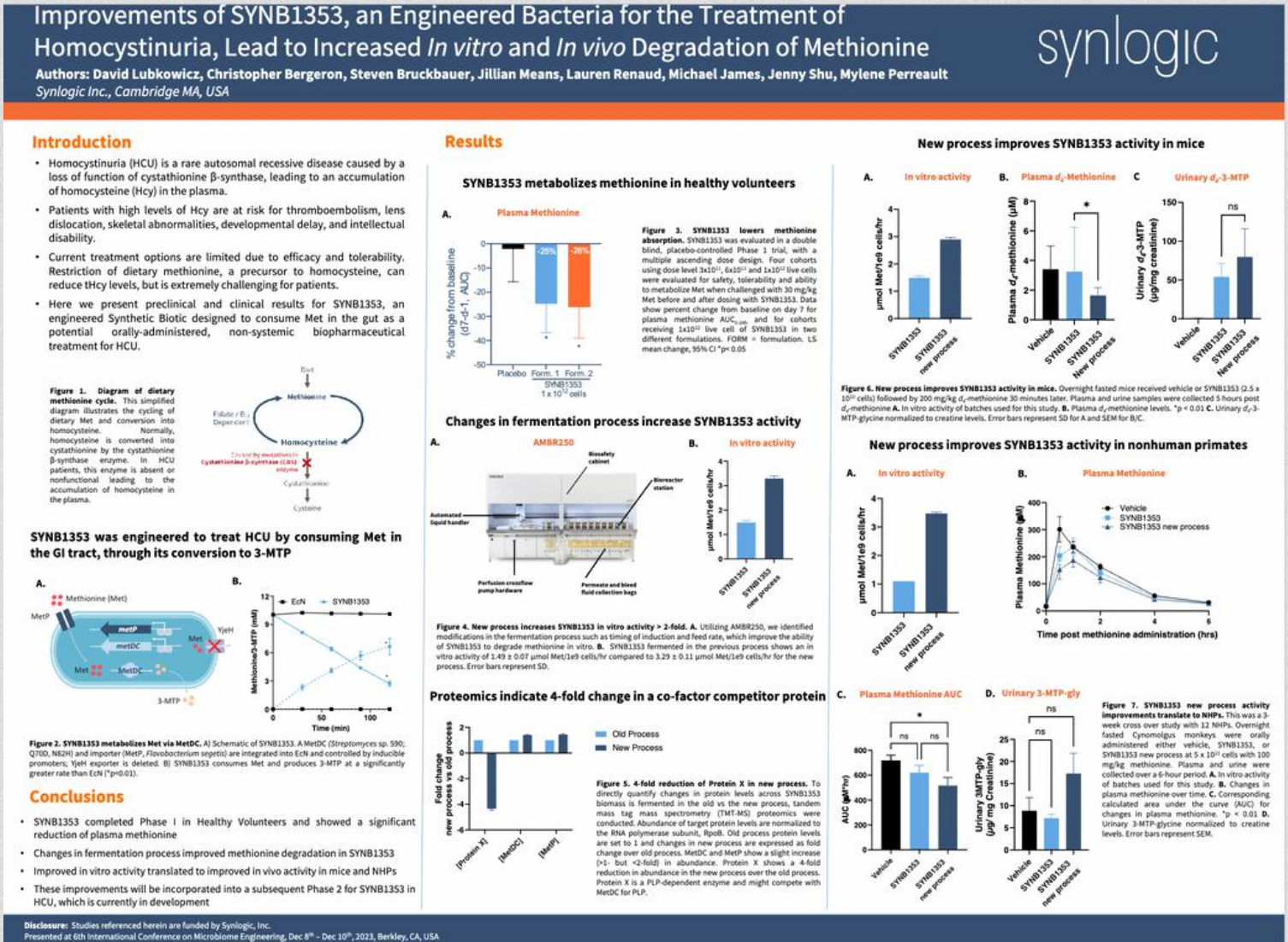
INDUSTRY NEWS

Synlogic Announces Abstract Accepted for Poster Presentation at the International Conference on Microbiome Engineering 2023

Poster presentation on SYNB1353 program for homocystinuria includes findings from process improvements to improve activity of methionine degradation

CAMBRIDGE, Mass., Dec. 07, 2023 (GLOBE NEWSWIRE) -- Synlogic, Inc. (Nasdaq: SYBX), a clinical-stage biotechnology company advancing novel, oral, non-systemically absorbed biotherapeutics to transform the care of serious diseases, announced that an abstract outlining the SYNB1353 program as a potential treatment for homocystinuria (HCU), including findings from fermentation process improvements to increase activity of methionine degradation, has been accepted and will be presented at the International Conference on Microbiome Engineering 2023, held in Berkeley, California on December 8th to 10th.

To read the full press release, visit: https://bit.ly/4aeAsHX



INDUSTRY NEWS

Travere Therapeutics Initiates Pivotal Phase 3 Clinical Trial of Pegtibatinase for the Treatment of Classical Homocystinuria (HCU)

Potential for pegtibatinase to become first disease-modifying treatment for classical HCU; topline data expected in 2026

SAN DIEGO, Dec. 14, 2023 (GLOBE NEWSWIRE) -- Travere Therapeutics, Inc. (Nasdaq: TVTX) today announced the Company has opened enrollment in the HARMONY Study, a global, randomized pivotal Phase 3 clinical trial of pegtibatinase, a novel investigational enzyme replacement therapy being evaluated for the treatment of classical homocystinuria (HCU). Classical HCU is a rare genetic metabolic disorder caused by a deficiency in the enzyme cystathionine beta synthase (CBS). The study is designed to determine the safety and efficacy of pegtibatinase in reducing plasma total homocysteine (tHcy) levels, a key treatment goal in classical HCU, compared to placebo in participants who are receiving standard of care.

To read the full press release, visit: https://bit.ly/48hgSsx

TRAVERE™ THERAPEUTICS

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

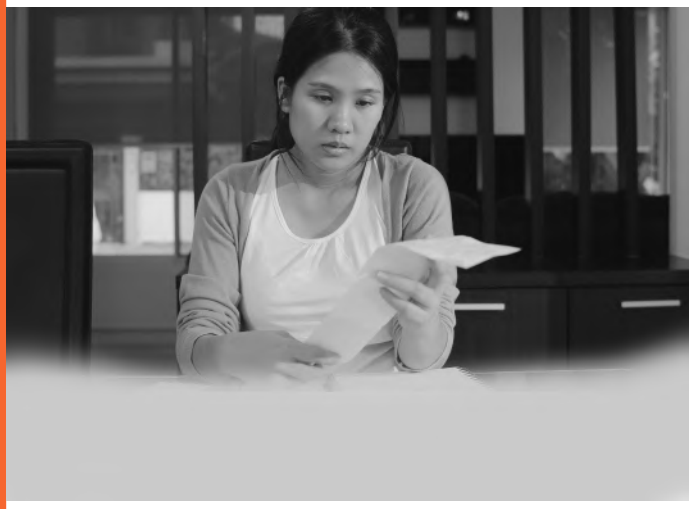
Monday-Thursday 8:30am – 7:00pm ET
Friday 8:30 am – 6:00pm ET

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:
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**

FOLLOW US