The HCU Herald

Featuring...

September is Newborn Screening Awareness month!

HCU Hero
Rileigh from Ohio

HCU NETWORK AMERICA

September 2023

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

HCU HERO:
RILEIGH FROM OHIO

UPDATE

5 year update Edition!

In 2019, when Rileigh was 19 months old, her family shared her HCU Hero story with us for the first time. You can read her original story here.

Keep reading to find out how Rileigh is doing today!

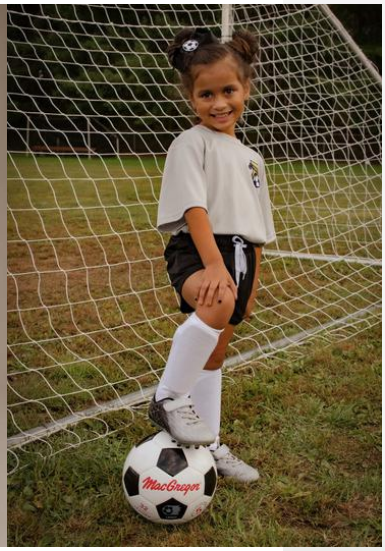
Rileigh was born a little over five years ago, on June 18th, 2018. Two days after she was born, and before packing up to take our newborn home, our hospital did a routine newborn screen that tests for 30 different conditions. After being home for a couple of days, I received a call from our hospital stating that Rileigh's amino acid test came back abnormal and needed to be retested. Instant worry set in. We headed to the hospital the next day to have her labs redone. A few days later, I got a call from our pediatrician telling me that they had set up an appointment for Rileigh at Nationwide Children's Hospital's genetics department. At that moment, I went straight to the internet, something I wish I had never done, and my heart sank. In an effort to prepare myself and my partner as to what we were up against, I read about all the worst-case scenarios possible, but it just sent me into a state of panic, and fear of the unknown.

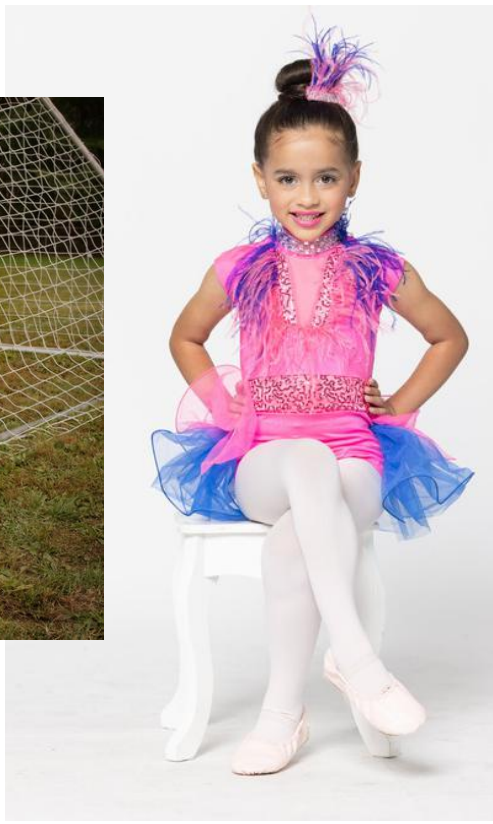
Rileigh was 2 weeks old during her first visit with the geneticist. I was in tears the whole visit, but he assured me that she was going to be fine as long as I followed his strict directions and adhered to the treatment that Rileigh would need. It all sounded so overwhelming. We went home and started treatment right away. It consisted of Hominex formula mixed with regular infant formula, Cystadane powder, folic acid and B6. Routine lab work and clinic visits were our norm during that first year of her life. Her lab work looked good each time which was always a relief. As a parent trying to navigate through caring for a child with a rare disorder, you're always wondering if you're doing everything right, and getting good labs always helped to reassure me.

Rileigh's first year was such a blur. I was always praying that she would grow and develop normally. Once she started solids, we went into another overwhelming phase, which was navigating the low-protein diet and making sure that we stayed within the 9 grams of protein a day that Rileigh was allotted. Baby food seemed so much easier than regular food, and we figured it out. We used a whiteboard for tracking the formula and food that Rileigh consumed each day, which we still use today (she's still on 9 grams of protein a day, too). She even likes helping to add up her protein amount and moving her magnet up until she reaches her goal. Rileigh also loves helping to prep her meals, like with her favorite vegetable soup! We keep her very involved with her diet and teach her the importance of her diet and medication for her health.

We now do yearly appointments with her geneticist, and we've recently had her 5th year check up! Rileigh is a very smart, energetic, happy child. She's been in tumbling since age 2 and has been involved in swimming lessons, soccer, t-ball and dance, too! (Her favorites so far are tumbling and dance.) We never want her to be limited by her disorder, and we tell her that she can do anything she puts her mind to.

Rileigh will be starting kindergarten this fall and we are a little nervous, but we've spoken with her school about her dietary needs and they've assured us that they would follow her strict requirements. We will pack her lunch and snacks, and she will drink her daily formula with the school nurse. This is just our next chapter of our little HCU child's life! She's doing so well, and I have to give so much thanks to her doctors, nurses and medical team! They know what they're doing. I also have to give my husband and I a pat on our backs as well. We were so scared and overwhelmed in the beginning, but we've managed very well and have become a great team. I know we'll inevitably face some challenges in the future, but right now we're all three feeling like rock stars!





To read Rileigh's 5-year update story on our website, visit
https://hcunetworkamerica.org/rileigh-2/

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!

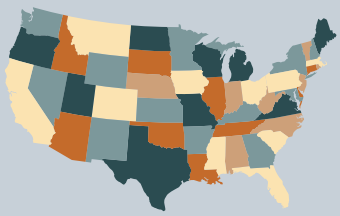
Email us:
dbartke@hcunetworkamerica.org

Newborn Screening Awareness Month

Celebrating 60 years!

Click here to see a *History of Newborn Screening* Timeline!

Visit Baby's First Test to learn about what conditions are screened for in YOUR state!



What can I do to advocate & spread awareness for Newborn Screening?

Share your story!

Whether it's in person or on social media, sharing your own experience with Newborn Screening is a great way to spread awareness!

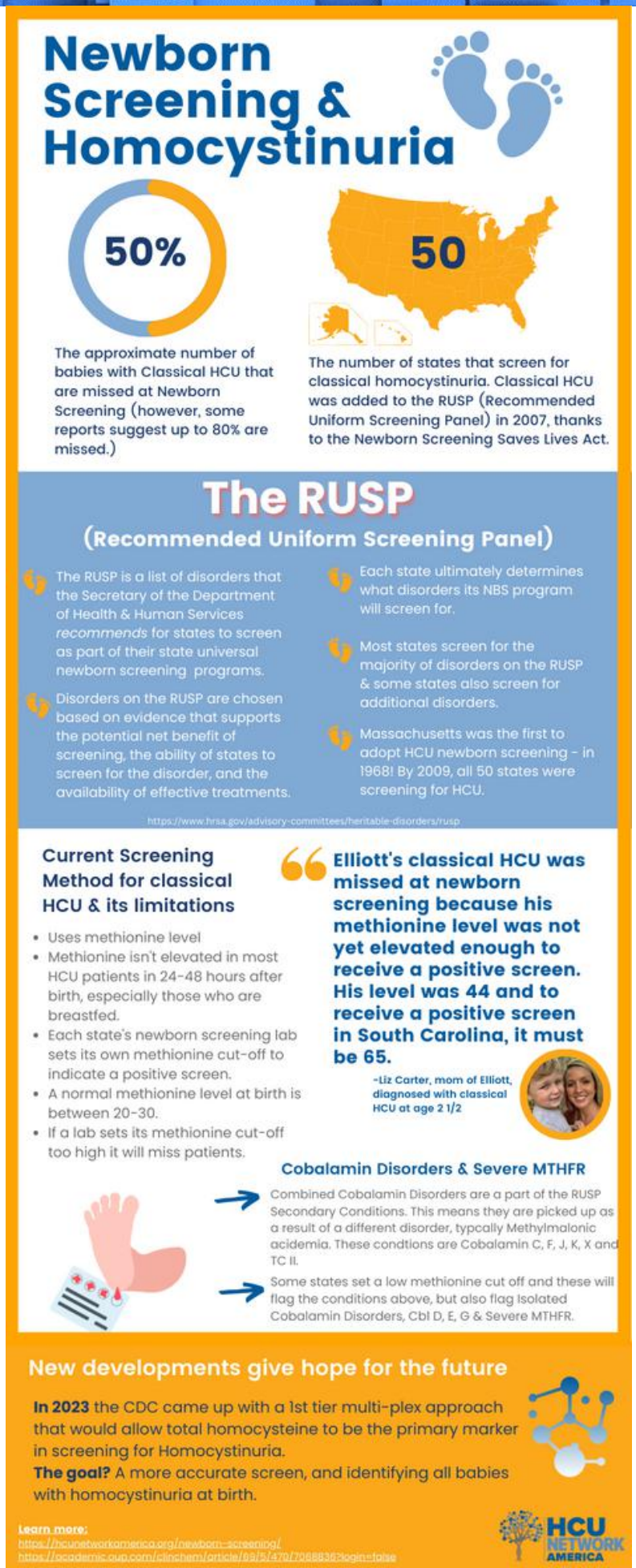
Leverage social media

Like, comment & share posts from advocacy organizations like Expecting Health, EveryLife Foundation, NORD & of course, HCU Network America! *(you can also search #NewbornScreening to find more posts to share!)*

Connect with advocacy organizations & sign up to attend events!

Rare Disease Week Events are a good place to start! (EveryLife Foundation/RDLA's Rare Disease Week on Capitol Hill, Rare Across America, or YARR program, for those ages 16-30!)

Newborn Screening Awareness Month



CHECK OUT

our new infographic!

Click here to download

Advocacy idea!

- Share this infographic, along with something about your newborn screening story to social media in September for Newborn Screening Awareness Month!
- Don't forget to use the hashtags:
  #NewbornScreening
  #2023NBS
  #NBS60years

UPCOMING EVENTS

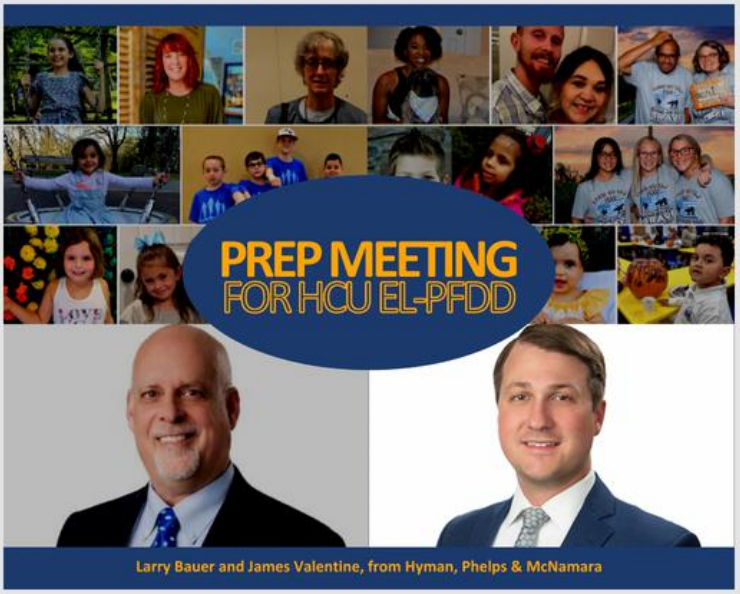
Register for these events at: https://hcunetworkamerica.org/virtual-meet-ups/

Prep Meeting for HCU EL-PFDD

September 28 at 4 pm ET

Hear from Larry Bauer and James Valentine, from Hyman, Phelps & McNamara. In private practice, James and Larry have worked with many patient organizations to ensure their community's voices were heard by decision-makers. Relevant to our EL-PFDD meeting, they have been involved in helping plan and moderating three-fourths of the over 75 externally-led PFDD meetings.

The Community Prep Meeting for the HCU EL-PFDD is an opportunity for everyone in our community to learn more about the upcoming EL-PFDD for people living with HCU.



We will discuss the following topics:

- What are the phases of drug development and how is the FDA involved at each step
- How does the FDA incorporate the patient and caregiver voice into their review of new clinical trials and in their review of new medical products
- Why is this meeting so important for the HCU community
- How can people from the HCU community participate in the meeting and help this meeting be a great success
- What are the topics most important for the FDA to learn about HCU

GET READY FOR HCU AWARENESS MONTH!

WEAR YOUR AWARENESS

October is HCU Awareness Month!
Get your Gear NOW!

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

Awareness Month Activities

- ☐ 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 #ToastTocHCU
- ☐ 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/

Save the Date!!!

...for our 2024 Family conference!

June 29-30th 2024
Aurora, Colorado

HCU NETWORK AMERICA

- 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!

Registration opening in October!

HCU NETWORK AMERICA

Your Participation Shapes Our Future!

The future of Homocystinuria depends on you, the patient! There are a few ways to participate and help acclerate research and advance potential new therapies. Each one is unique and has benefits to the community. Before you join any kind of study, it's important that you understand what is expected of you and how it might give back to the community.

	HCU Data Collection Progam Powered by RARE-X	Natural History Studies (NHS) Classical: https://bit.ly/3WUgYQS Cobalamin: https://bit.ly/CblNHS	Clinical Trial	Market Research
What programs are available for my participation?				
Who collects my information?	Patient collected	Medical Team collected	Medical Team collected	A market research agency hired by a pharmaceutical company
Who submits my information?	Self-reported by patient or caregiver	Physician reported	Medical Team reported	Survey taker or self-reported by patient
What type of information is needed and how is it reported?	Caregiver or patient diagnosed with specific condition self-report their actual experience and current medical care from their home computer.	Physicians track the "natural history", or course a disease takes in individuals over time, beginning with the onset of symptoms and taking into account the patient experience with current treatment.	A volunteer patient follows a medical protocol, while physicians and nurses track the safety and effectiveness of all treatments or medical procedures.	Qualifying volunteer patients answer a series of questions about their actual experience and current medical care. Either they self-report or the survey taker submits responses.
What happens to my information and data after it's reported?	De-identified data shared by patients/ caregivers can be accessed by researchers for disease studies and clinical trial recruitment.	While no new drug is administered, a patient's collected information is essential for research, drug development, and clinical study design.	Clinical trials are the primary source for researchers to learn if a new drug or medical device is safe and effective for patients.	Data is analyzed to find insights and trends and to aide research sponsors in making informed decisions about their next steps.

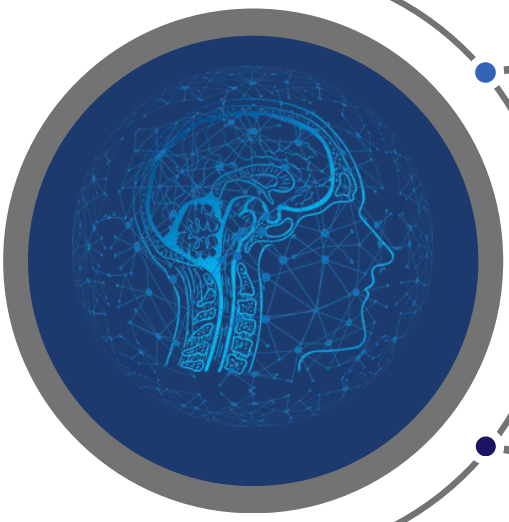
HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

Brain and Nervous System Survey

While some of these issues are better documented, other aspects aren't.

- Cognitive Impairment
- Coordination Issues
- Seizures
- Failure to Thrive

What are we missing? Help us connect the dots!



01 **Enroll at https://homocystinuria.rare-x.org/** (5 minutes)

02 **Take the Health and Development Survey** (15 minutes)

03 **Complete the Brain and Nervous System Survey**

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

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

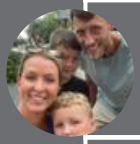
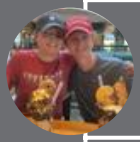
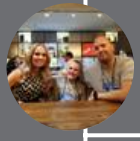
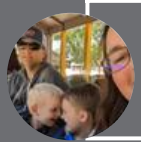
TRAVERE THERAPEUTICS

HCU HEROES RACE FOR RESEARCH

WALK / RUN / RIDE

HEROES IN THE MAKING

Meet our Teams / Click to read each team's story & donate!

 ***Amy's Army***	https://bit.ly/45jhAEO
Brooke's Blazers	https://bit.ly/3E1BesK
 ***Cakes for Carson***	https://bit.ly/45jbcgE
 ***Carter Crew 4 HCU***	https://bit.ly/44fMkVJ
 ***Ellie's Entourage***	https://bit.ly/3YG9qng
 ***Grayson's Gang***	https://bit.ly/45xXA0l
 ***"Hunt" for research***	https://bit.ly/45BxDO0
 ***Masen's Mad Dawgs***	https://bit.ly/3QMpvpz
 ***Mighty Marchese's***	https://bit.ly/3KNjZPO
 ***Race with C & G***	https://bit.ly/3QM4xqY

Recordati - Sylvia's Supporters	https://bit.ly/3sfsYTi
 ***Renna's Rare Runners***	https://bit.ly/3E1Qy8T
Synlogic	https://bit.ly/3sepTmp
 ***Team Anniston***	https://bit.ly/3P3mcZT
Team Eton	https://bit.ly/3E0E15A
Team Hawkins/PwC	https://bit.ly/3QQtlOp
Team Recordati	https://bit.ly/47HcRhu
 ***The Bartke Ruff Ruffs***	https://bit.ly/47H1Oou

Follow each team's progress throughout the month by visiting our race website!

https://charity.pledgeit.org/HCURaceforResearch

HCU HEROES

RACE FOR RESEARCH

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

HCU NETWORK AMERICA

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Finisher sponsor

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Cheer Section Sponsors

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synlogic

Learn more about Synlogic

UPDATE

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

NORD®

CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

What is the purpose of this program?

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

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

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

MEDICAL ASSISTANCE

Who is eligible to apply?

This program is designed to help patients who:

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

What is the application process?

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

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

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

NORD is Here for You

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

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

Alone we are rare. Together we are strong.®

How do I get more information and apply?

Contact NORD's Classical HCU Medical Assistance Program

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

203-616-4327

203-635-4163

hcu@rarediseases.org

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

What assistance does NORD provide?

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

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

Awards are issued for a calendar year.

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

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

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

How does NORD demonstrate compliance with regulations required of charities?

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

NORD®

rarediseases.org

Rice Pudding

Makes 5 servings | Serving Size 1 cup | .5 g protein per serving or 21 mg PHE

Ingredients:

- 168 g Low Protein Rice, about 1 cup
- 3 c Rice milk, divided
- 1/2 tsp Salt
- 1/2 tsp Cinnamon
- 1/4 c Brown Sugar
- 1/2 tsp Vanilla Extract
- 1/4 tsp Ground nutmeg

Directions:

1. Bring 2 cups of rice milk (or any non-dairy milk) to a simmer over medium heat. Whisk occasionally to prevent the milk from scorching.

2. Add the low-protein rice, salt, cinnamon, sugar, and vanilla. Stir to combine and dissolve the sugar. Allow to cook, stirring frequently to prevent burning. Add more rice milk as the rice begins to thicken as needed. Cook until rice is soft (about 30-40 minutes.) Allow to set for 5 minutes before serving to thicken. Sprinkle with nutmeg and serve warm.

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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