The HCU Herald

Featuring...

HCU Hero
Matteo from Delaware

July 2024

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

HCU HERO: MATTEO FROM DELAWARE

Introduction:
In April 2021, just 9 months postpartum with my daughter Mia, I was surprised to learn of my second pregnancy. We were overjoyed to discover we were having a son! This is the story of our journey with Matteo, from the early days of joy to the challenges we faced with his diagnosis of Cobalamin C Disorder (CblC).

Pregnancy and Birth:

Having been in the fitness industry for the last 14 years, I stayed active and maintained a healthy diet throughout my pregnancy. I was much smaller in comparison to my first pregnancy but chalked it up to my hard work through fitness. Since I had just had my daughter in 2020, and it was less than a year later, in 2021 I opted not to have genetic testing of any sort done. How much would have changed in 9-10 months, right? I felt my pregnancy was great! I had lots of energy, and everything seemed normal to me. Matteo was born on a Sunday in December at 39 weeks. It was a smooth, easy delivery, and he seemed perfect in every way. He was a peanut at just 6 pounds, smaller than our first child who was 8 pounds 2 ounces. We received no indication that anything was wrong from the delivery team.

Initial Concerns:
Matteo slept a lot in the recovery room, longer than I expected even for a newborn, and he didn't want to eat much in his first 48 hours. We thought it was minor jaundice, like what our daughter experienced, so we weren't alarmed. We stayed in

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the hospital for 48 hours. Matteo was put under the “baby lights” and we were sent home with some instructions to see the pediatrician about the jaundice levels. We still had zero indication that anything was wrong. However, on our first night home, Matteo slept overnight from 9 pm to almost 6-7 am. I was terrified when we woke up, as a crying baby hadn't woken me up. Despite reassurances from family that he was just a "good baby," my gut felt something was off. I began to make calls to the pediatrician and ER.

Diagnosis:

Not even a few hours later, we received a call from the hospital telling us that some elevated levels had come up on Matteo’s newborn screening. We were panicked. I distinctly remember hearing the phrase “urgent but not emergent.” They gave very little details, but all of my fears over the last 48 hours were confirmed. I called Matteo’s dad crying hysterically. We were instructed to go to Washington DC, a drive more than 2-hours away. We had a 1 ½ year old toddler, and I didn’t yet fully understand what was happening. I advocated going to the metabolism team at CHOP in PA which was about 35 minutes from us instead. Just 20 minutes later, I got a call from the on-call doctor at CHOP telling us Matteo had what they thought was Cobalamin C and needed to come to the ER for confirmation and bloodwork. I remember listening to the doctor on the speakerphone, while sobbing and looking at my beautiful new baby, scared of what was to come.

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At the ER, Matteo was hooked up to machines and IVs. Several tests were done, and we received vague instructions while waiting to meet the metabolism team in person. The voices from that night still swirl in my head: " *You will need to change to breast milk as soon as possible* ", " *You will likely need to give him a shot every day.*" , " *Protein of all kinds will be a problem.* ", " *Do you know much about b12?* ", " *He may have heart, eye, and muscle issues in the future.*" , and " *We don't know what his neurological development will be like.* " What did any of this mean? I was too stunned to ask questions, nor did I know which ones to ask. The next day, we moved from the ER to the metabolic wing and received the confirmed diagnosis of Cobalamin C Disorder. Our family drove home with our new diagnosis, some syringes, some syrupy meds, and a new way of life we didn't know how we were going to navigate.

Early Challenges:

That night, I took to the internet. In 2021 without having a Facebook account, I found that there wasn't much out there but extremely difficult-to-read articles. TikTok had nothing and Instagram had limited others sharing their stories. We had no idea what we were in for. I started trying to learn anything and everything I could. We felt scared, overwhelmed, and shocked. We had no tribe to get us through. We did so much on our own in terms of Matteo's medical advocating, learning, and treatment.

Cobalamin C (CblC) deficiency is a rare, inherited metabolic error that prevents the body from processing vitamin B12 properly. It's caused by mutations in the MMACHC gene and can affect people of all races and ethnicities. CblC deficiency can present in children or adults, but it can be difficult to diagnose in adults because of its rarity and many possible symptoms. Early diagnosis can help prevent permanent neurological damage.

Now, when I tell people about our story, (learning of the diagnosis and being newly postpartum) I always feel like this part was a dream, and then I blacked out for about the next 12 weeks.

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One of the earliest memories of Matteo's diagnosis was having our metabolic specialist doctor drive medication to our home to see Matteo through the doorway (it was during COVID) and to make sure that ALL of us were ok. It was one of the most heartwarming, incredible moments I have felt. The first few weeks were difficult as we adjusted to giving shots and meds and starting to breastfeed and pump, all while caring for two babies. We compartmentalized our worries and didn't share much about Matteo's diagnosis publicly for fear of not having the answer or becoming emotional. I frequently called the doctors with questions, and they were extremely helpful, sensing my highly emotional and scared state. Everyone in our family had an opinion they found on the internet over the next few weeks, and it was overwhelming to decipher. Over the next 6 months, we saw CblC manifest in various ways. Matteo was in the 2nd percentile for height and weight, had low muscle tone, had trouble tracking with his eyes, and was extremely sleepy. He didn't learn to sit up until he was nearly 8 months old, and it was with the help of an amazing physical therapist who we worked with until Matteo was roughly 14 months old that he could stand on his own. Matteo would have good days and bad with fatigue. Overall, we dealt with each day with strength and knowing that Matteo was going to be the best version we could give him.

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Major Health Scare:

When Matteo was 4 months old, he contracted COVID-19. It was one of the scariest parts of our journey. He began to stop eating, was losing weight rapidly, was coughing, sleeping more, and not taking his daily meds. Getting an infant to take oral meds was hard, let alone while having

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an illness. Matteo was hospitalized, hooked up to IVs, EKG, and pulse oximetry. I remember looking at him and seeing how incredibly tired he looked and PLEADING with the team to find a way to help him. The next morning when he woke up, after good fluids and meds, Matteo was a little more like himself again and for the first time in 3 days, I could breathe. It was then I learned that this would be our life: on the constant edge of quick decisions and hospital visits whenever Matteo contracted a severe illness. This would be the first of many hospital stays for us.

Ongoing Management:

Matteo, who we fondly call *Teo Bob*, is now a thriving 2 ½-year-old who defies the odds of his rare disease each day. He attends a medical daycare program where he receives occupational, physical, and speech therapy daily—something I wish we had started sooner. Matteo is as mischievous as any little boy, loves basketball, baseball, and football, and enjoys snuggling, reading books, and playing with his sister in the evenings.

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Reflection and Advice:

In the 2 ½ years since Matteo's diagnosis, we have experienced a rollercoaster of emotions. We celebrate the small wins, take each day as it comes, and learn about the challenges that lie ahead. We rely on our friends in HCU Network America as our tribe when we need help. Explaining his rare disease to others, I feel passionate and excited to raise awareness. Every day feels like overcoming a new challenge, from hunting down out-of-stock meds to balancing nutritious meals and teaching Matteo new words. My advice to anyone who has just received a diagnosis is: find your people, extend your network, ask for help, and advocate hard. No one knows your child like you do. In the future, we hope to learn more about ways to help Matteo's development, that his symptoms don't present themselves harshly, and that he gets to live the life that he dreams!

Click here to read Matteo's & other patient stories on our website!

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

CONFERENCE RECAP!

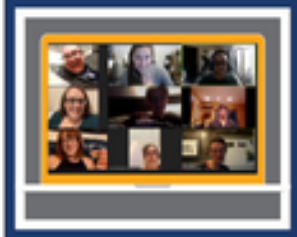
Thank you to all who attended our 2024 Moving Mountains Family Conference in Aurora, Colorado!

Keep an eye on our social media for photos & videos of the conference content!

Click to follow us!

UPCOMING EVENTS

Our virtual meetups are an opportunity for patients & caregivers with any of the homocystinurias to connect, learn, and share experiences. We will also share important community updates!



https://bit.ly/summer-meetup

MEET OUR NEW MEDICAL ADVISOR!

Walla Al-Hertani,
MD, MSc, FRCPC, FCCMG, FACMG

Dr. Walla Al-Hertani is a Biochemical Geneticist and Clinical Geneticist with expertise in inborn errors of metabolism (IEMs) and a special interest in Lysosomal Storage Diseases. Dr. Al-Hertani received her Doctor of Medicine degree from Dalhousie University in Halifax, Nova Scotia, followed by the completion of a Clinical Genetics residency at the University of Ottawa and a Biochemical Genetics fellowship at the Hospital for Sick Children (SickKids) and the University of Toronto. Dr. Al-Hertani practiced as a Medical Biochemical Geneticist and a Clinical Geneticist at the Montreal Children's Hospital, and McGill University, followed by the Alberta Children's Hospital in Calgary, Canada before relocating to Boston, where she was the Harvey Levy Endowed Chair in Metabolism and served as the Medical Director of the Metabolism Program, the Lysosomal Disorders (BoLD) Program and the Glycogen Storage Disorders Program. She is actively involved in a number of clinical trials investigating innovative therapies for IEMs and rare diseases. Dr. Walla Al-Hertani is currently the Division Chief, Metabolic Disorders Division at the Children's Hospital of Orange County.

UPCOMING EVENTS

EVERYLIFE FOUNDATION FOR RARE DISEASES

15TH ANNIVERSARY

RARE ACROSS AMERICA

EVERY VOICE, IN EVERY DISTRICT, MATTERS

AUGUST 5-16 2024

Registration for Rare Across America is open until Friday, July 12th

Rare Across America 2024 is the opportunity to meet with your Members of Congress at their in-district offices and educate them on the issues that are most important to the rare community by sharing your story.

Where?

All Senate meetings will be scheduled virtually and House meetings will be in person at your Member's in-state, in-district office. Rare Disease Legislative Advocates (RDLA) will schedule meetings for you and help you to prepare. No prior advocacy experience is necessary.

When?

Meetings with Members of Congress will take place between August 5th and August 16th. We will be offering training webinars to help advocates prepare for their meetings.

July 16th, 12pm ET: General Training Webinar – Register Here
July 24th, 12pm ET: Team Coordinator Training Webinar (A link to attend this training will be sent to all confirmed team coordinators).
July 25th, 12pm ET: Share Your Story with Policymakers Webinar – Register Here

REGISTER HERE

UPCOMING EVENTS

EVERYLIFE FOUNDATION FOR RARE DISEASES

15TH ANNIVERSARY

2024 NEWBORN SCREENING BOOTCAMP

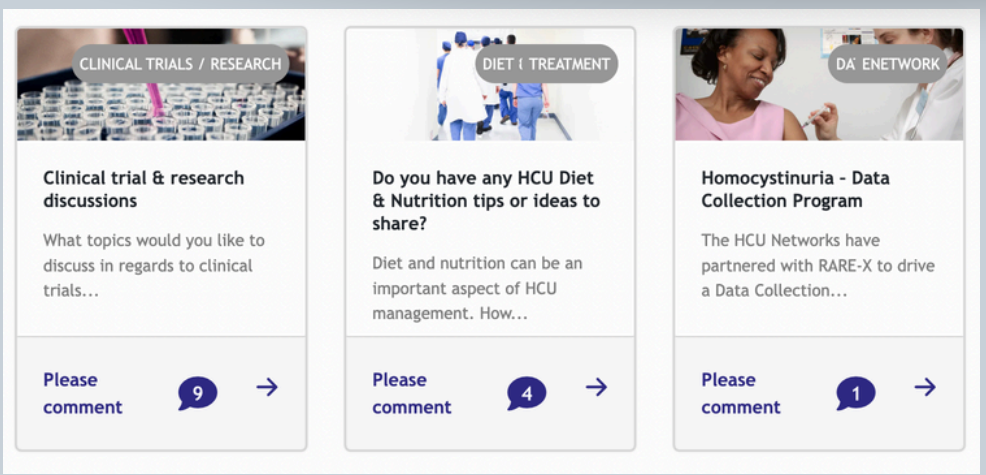
REGISTER HERE

JOIN THE HCU E-NETWORK!

Welcome to the HCU eNetwork

Powered by HCU Network America & HCU Network Australia, we aim to utilize this platform to connect with HCU patients and carers worldwide and gather your input on key topics in relation to HCU diagnosis, management and treatment.

Questions and activities will be updated on the platform throughout the year, so please check back regularly and look out for email communications that will be sent out notifying you when new topics are posted.



HCU NETWORK AMERICA

HCU Network Australia

Join the conversation!

https://hcuenetwork.org/

2023 Featured Race Teams

Team Hunt

$16,146 raised for Cobalamin G research!

Cakes for Carson

$4,540 raised for Severe MTHFR research!

Ellie's Entourage

$2,425 raised for classical HCU research!

Check out next month's Herald for more featured race teams from 2023!

Cystadane; the last FDA approved drug for the homocystinurias (HCU) was approved in 1996. That was 28 years ago!

Currently is only one clinical trial in progress for classical HCU, but there are none in progress for cobalamin disorders and Severe MTHFR.

In collaboration with other organizations, HCU Network America has issued 7 research grants.

Help us accelerate better treatments for and help raise funds during our Race for Research!

https://bit.ly/HCURace24

RECORDATI RARE DISEASES
Focused on the Few®

Pricing

- **$5 - no shirt or medal, participate and fundraise!**
- **$20 Medal or shirt only**
- **$30 Both!**

Incase you missed it...

The HC&U Podcast is back!!!

HC&U is a podcast about Homocystinuria, sponsored by HCU Network America and hosted by Ben & Lindsey.

Meet your hosts!

Welcome to the HC&U Podcast! We are Ben and Lindsey, your hosts. We are so excited to be starting this as extra resources for the Homocystinuria community. We hope you like our content!

To Listen:

https://hcunetworkamerica.org/hcu-podcast/

or click below on your favorite option!

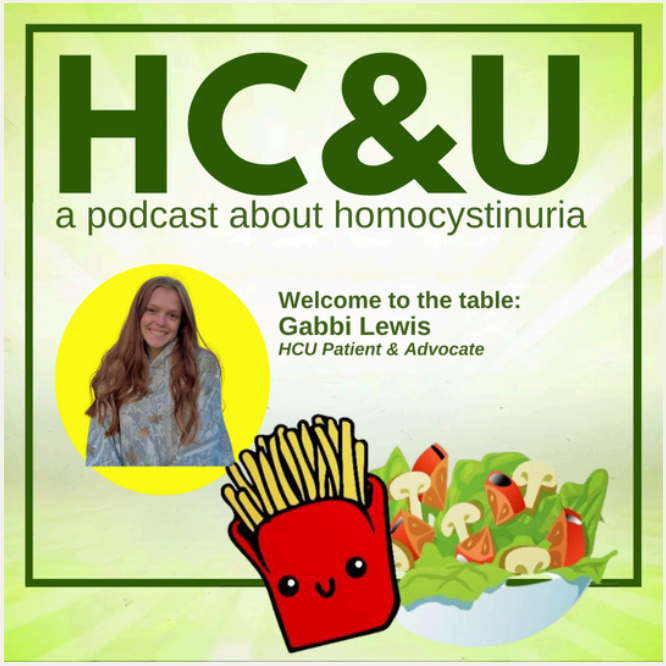
LISTEN ON Spotify

Listen on Apple Podcasts

LISTEN ON iHeartRADIO

Listen on amazon music

The latest episode



In this episode, Ben welcomes Gabbi to the table!

A recent college grad (not to mention a Double Major Summa Cum Laude!), Gabbi was the first baby diagnosed in utero with classical HCU. Despite early treatment, she's had multiple surgeries to address scoliosis and she and Ben will discuss just how these issues have impacted her, but also the incredible successes that she's found.

UPCOMING EVENTS

GREAT OAKS RETREAT CENTER

ILLINOIS LOW PROTEIN FAMILY CAMP

REGISTER NOW!

Check-in: 5:30 pm, August 9, 2024 | Check-out: 10 am, August 11, 2024
Great Oaks Retreat Center
1380 County Rd 900 N, Lacon, IL 61540

LODGING

Lodging is included. Please bring your own pillows and linens - blankets or sleeping bags. Beds are bunk beds in a large room.

WHAT TO BRING:

- Formula
- Sleeping bag or linens
- Favorite snacks
- Towel
- Bathing suite
- Sunscreen
- Bug spray
- Clothes to camp in
- Closed toe shoes
- Shampoo & soap
- Tooth paste & toothbrush
- Camera
- Phone charger

FRIDAY 08/09	Potluck Dinner Arts and crafts Bonfire and smores		
SATURDAY 08/10	Breakfast Tie Dye Group Photo Archery	Open swim Boating Fishing Lunch Ziplining/high ropes	Dinner Ghost in the Graveyard Bonfire and Smores
SUNDAY 08/11	Breakfast Closing Activity Clean and pack up	REGISTER NOW!	

REGISTER BY JULY 19!

https://bit.ly/3TKWdJc

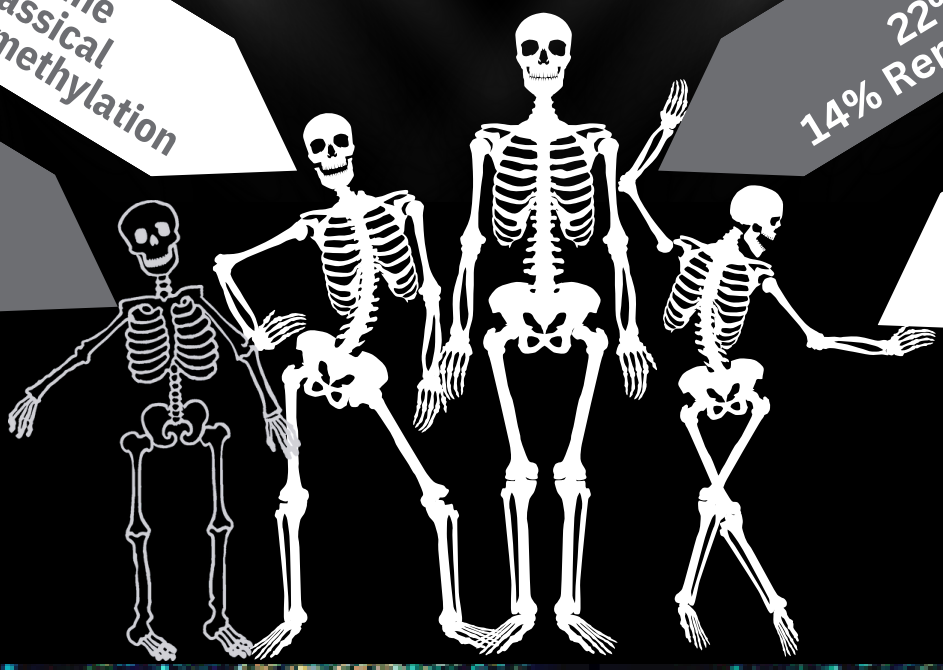
HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

POWERED BY RAREX

Spotlight on Bone, Cartilage, and Connective Tissue Health!

Curved spine
28% Classical
19% Remethylation

Osteoporosis
10% Classical
0% Remethylation



Tall Stature
22% Classical
14% Remethylation

Flexible Joints
15% Classical
6% Remethylation

Complete the survey

CANPCU+ CANPKU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RAREX

Now Enrolling

Acappella

Sponsor: Travere Therapeutics
Study type: Natural History (no investigational medicine given)
Study duration: About 6.5 years
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)	Currently enrolling 1 to 4 years old	The study will include three key stages (screening, enrollment, and observational follow-up) and will last approximately 6.5 years.

**Your child will need to meet all other study criteria to take part in the ACAPPELLA Study.*

Study Locations
United States: Colorado, Washington DC, Georgia, Pennsylvania
Countries outside of the US: Ireland and Qatar

Approximately 150 people aged between 1 and 65 will participate at sites in the US, Europe and other countries around the world.

150

The ACAPPELLA Study has already enrolled 100 adults and children over 5 years old, and is now looking for children aged 1 to 4 years old to take part.

You may be able to receive payment for time and travel if your child participates in this study.

Talk to your doctor and family members about your child 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

If you have any questions, please email:

medinfo@travere.com

For more information, please scan the QR code or visit:

www.hcuconnection.com.

MA-PE-24-0008. Version 2.0; en-US; March 2024

TRAVERE THERAPEUTICS

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!

Get your kit!

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*!

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

Stay connected: Join our Contact Register!

What is a contact register?

- a secured private survey that allows you to share information on you or your family member with HCU with us. (general contact info, diagnosis, etc)
- kept confidential and will not be shared unless you permit us

Why join?

- subscribe to our monthly newsletter and other communications
- identify other patients in your state and request their contact information
- access information posted over time that can only be shared with the patient community
- helps us plan events and inform the development of resources and educational tools
- supports our advocacy efforts and enables us to have informed conversations with doctors, pharmaceutical companies, state newborn screening labs, and lawmakers.

I want to participate! What's next?

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

HOPE CONNECTS US

FOLLOW US