The HCU Herald

Featuring...

HCU Hero
Marley from Texas

August 2024

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

HCU HERO: MARLEY FROM TEXAS

We found out that we were pregnant with Marlene "Marley" right around 3 weeks. I remember telling my husband, Dalton, around 5 weeks that I wanted to wait to tell other people. I had a feeling that I might miscarry or that something could be wrong; not because of anything physical, just an inner intuition that I felt. I could hardly explain it. Everything ended up being fine, and I had an overall easy pregnancy, similar to my first pregnancy with Marlene's older sister, Addy. About two months before Marley was due, her ultrasound showed that she had gone from the overall 10th percentile in all areas, down to the 3rd percentile in her tummy area. This led to a diagnosis of IUGR (Intrauterine Growth Restriction) and required weekly ultrasounds. She did jump back up to the 8th percentile in her tummy, but we ultimately decided to take her via planned c-section at 37 weeks and 5 days. My doctor decided at that point that she would get more nutrition outside of the womb than in. We agreed, and ended up birthing a beautiful and healthy 5lb 12oz baby girl! She was tiny, but looked great.

For the first two weeks of life, Marley's body temperature fluctuated. She lived on a heating pad, and in everyone's arms, all bundled up. Her "Pops" held her on the heating pad for 10 days, and "Lolli" chased big sister around. Her "Dandie" and "Poppy" saved us with snacks to refuel and help in any area we needed! We couldn't have done those first two weeks without any of them.

We were at the pediatrician's office every few days to check her weight, temperature, etc, and we couldn't have asked for a better or more supportive doctor and group of ladies in the office. At this point, we all thought she was just tiny and needed to get some more fat on her bones to maintain her temperature.

HCU HERO: MARLEY FROM TEXAS

She nursed great from the day she was born, so eating wasn't a struggle. We believed that at this rate, she would eventually put on more weight and maintain her temperature on her own.

At 14 days old, we went for a visit with her pediatrician and left with a plan to continue warming her up as needed, and monitoring things closely. But that same evening, we got a call from the pediatrician. We initially thought she was calling just to check on us, but the conversation quickly turned. She told us that a geneticist from Cook Children's Hospital had called her, and while Marley's first newborn screen wasn't flagged as out-of-range, her secondary screen showed some elevations. She went on to say that after chatting with the geneticist, they both agreed that there wasn't a need to rush her to the ER, but that we would need to take her in the next day for bloodwork. I remember hearing "This could be a fluke."

My husband Dalton and I felt strongly that something could be wrong with Marley, but truly had no idea what would unfold in the days ahead. We prayed for that fluke; that we were right in thinking that she just needed more time to put on weight and praying that time was all that our baby girl needed. We both agreed that there was no reason to rush, and that we should spend one more night together and take her in for labs in the morning.

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About an hour after we got off the phone with her doctor, Marley's temperature was down to 93°, and she was obtunded (worse than lethargic). Up to that point, she had been alert and responsive. We immediately warmed her back up on the heating pad as

HCU HERO: MARLEY FROM TEXAS

we grabbed what we could, and rushed to Cook Children's, our closest children's hospital. We got there at 8 pm and by 8:16 she was under a warmer, with all kinds of tests being run. I am still amazed by how quickly and efficiently the team worked. This is where our timeline to diagnosis started.

3/17/22

We were admitted to the NICU. Marley was administered an IV line and had lots of labs drawn, resulting in several blown veins. We met the geneticist that morning at 8 am, and she was wonderful. She listened as we told her all about Marley's first two weeks of life, and she took the time to explain Marley's newborn screen results to us. Since her first screen did not flag with major elevations, she told us that either we could be dealing with something very serious, or it truly could be an anomaly, and that the pending lab results would give us more information. Unfortunately, the labs had to be sent out, so our wait began.

3/18/22

As we waited for lab results, I was nursing and pumping, bonding with my new baby, and hoping my milk was good for her. I checked continually to see if labs came back and prayed that the results would be normal. I was really missing our oldest, Addy Rose, but remained hopeful we'd be going home soon.

03/19/22

This would be another day of waiting for lab results to come back, full of constant monitoring, worry, and not enough sleep. Dalton went home and packed our bags (since our stay was becoming longer than expected), loved on our dogs (thanks to our wonderful neighbors for helping with them!), returned to the hospital, and got me outside. We ate lunch, rocked on the

bench, and I fell asleep; I had needed the fresh air more than I realized. We were both exhausted physically and emotionally. Our sweet little baby continued unable to hold her temperature on her own but did eat very well. There was still so much unknown at this point!

3/20/22

Late the night before, one of Marley's labs came back. It showed an extremely elevated homocysteine level. It was 200, and we were told that the normal range is around 4-15. This led the team to believe that she either was deficient in b12, or that there was something more serious going on. They wanted to check and see if perhaps I was a b12 deficient (which would've been the best possible outcome), so they sent me off to get labs as well. Regardless of my pending results, they planned to come in at 9 am to begin to administer b12 injections to Marley. It was midnight when they informed me, so I texted my husband, overwhelmed but relieved that we had somewhat of an answer and plan. They basically told me that if she needed the b12, the injections could save her from going into a metabolic crisis, but that if she didn't need it, that it wouldn't hurt her.

Before talking to her geneticist that morning, I looked up what I could with the little information I had. The geneticist told us that we still had a spectrum of things to rule out; that Marley definitely had something deeper going on and the additional labs would help to narrow it down. She stressed the importance of continuing the b12 shots, as they could potentially prevent further harm. Marley was still eating so well but ended up losing 20 grams of weight that day. That night, she had bath time with one of our favorite nurses. Thankfully, pieces of the puzzle were slowly starting to come together, but there was still so much unknown.

3/21/22

A day of overwhelm and emotions! I was really missing our older daughter Addy, and I had to leave Marley for the first time since she was born to go get my labs drawn. It was nice to get out of the walls of the hospital, but very hard to leave my baby girl

for the first time. I also felt mom-guilt and wondered if everything was my fault. Did I have a b12 deficiency and just not know it? Was I not eating good enough while pregnant, and caused her IUGR (intrauterine growth restriction)? I had so many questions and so many emotions. Dalton and I went to get my labs done, grabbed a few things at CVS, picked up lunch, and then went back to the hospital.

Marley still wasn't gaining weight. At birth she weighed 5lb 12oz, but at this point, she had gotten down to 4lb 9oz. One of her doctors made me feel like I wasn't feeding her enough, which really put me in a hard place mentally. She was nursing great every time I fed her, the extra milk I was pumping was fatty, and I was building up a stockpile. We even had speech come in to do an evaluation, to make sure that this wasn't all caused by a feeding issue. Our lovely speech therapist watched her nurse and drink from a bottle. We tried several different ones, because she was a sloppy eater, and had a possible lip tie, but my supply and her latch were not an issue. She was definitely getting the milk she needed.

Later that day, the doctor on call looked over and assessed Marley's whole situation thus far. She said she thought her feeding was perfect and said her weight gain and body temperature issues could truly be due to her needing the b12. That morning was only her second shot of b12, and they originally said we'd hopefully see some improvements within a few days. At this point, we just needed more time to get results and see what her body was going to do. Deep down I felt that the b12 injections could be just what Marley needed. She was wiggling around some and was more vocal that evening, and she'd never been like that before. I wasn't sure if it was just me hoping and praying that it was helping, or if it actually was bringing her back to life.

In addition to all that was going on, Marley's geneticist was attending a virtual conference that week and shared that it would be her last week at the hospital; she was moving up north to be closer to family. We were sad to hear the news, but despite her moving away, she remained a presence in our lives. She called and talked

to me every single day. She wanted updates on how Marley was doing (from me personally!) and wanted Dalton and I to know that she was not going to leave us alone until she could give us some answers. Even today, it makes me want to cry thinking about it. She checked Marley's lab results every day, and even called the lab in Utah several times to bug them, because one of the labs had gotten lost. She was AMAZING! We were super sad to have met her and to be losing her all in the same week. She was a huge part of the beginning of our journey.

03/22/22

After sitting on it for a few days while waiting for lab results, and reaching out to another doctor, the geneticist brought her college textbook in, sat us down, and showed us exactly what she was thinking might be going on. She even drew us a picture, so that we could really visualize what she thought could be going on inside of Marley's cells. That day started our learning and teaching journey!

That night we added Betaine to her regimen, along with a few other medications. Her geneticist also ordered more labs to be drawn the next morning to determine if the b12 was working. Dalton and I knew it was indeed helping her, before any tests were even run; she was turning into a different kiddo! Marley also gained an ounce that day. It was like she was coming back to life right in front of us.

3/24/22

Marley's b12 lab came back and showed that she was not deficient, but her Methylmalonic acid (MMA) lab had a massive elevation. The MMA and homocysteine elevations ultimately showed, without yet having the genetic testing to confirm, that she had an inborn error of metabolism. Now we were most certainly looking at a metabolic disorder instead of just a b12 deficiency. This was one of the worst-case

scenarios that our geneticist first presented to us, but we were encouraged by the fact that she seemed to be responding well to the b12 injections. We didn't have proof that the meds were working yet, as we were waiting on her next set of labs, but we couldn't ignore the fact that Marley was starting to act like a true newborn baby who wiggled, made noises, and reacted to our voices with facial expressions.

3/25/22

This was another slow progress day, full of prayers that Marley would start gaining some weight and maintain her temperature without help from the warmer. She was not quite back up to birth weight yet but was starting to gain little by little. We were celebrating grams at this point!

3/26/22

That night, we weighed her three times, just to make sure we were getting it right. Her first weight was 2440g, and the second two were 2450g. We took 2450! Our nurse had no idea, but that number was extremely special to me. It is my Memaw and Pawpaw's house address. I knew that it had to have been Memaw giving us a little wink from Heaven, and I sure needed it that day!

One of Marley's repeat labs also came back that day. Her Homocysteine level had come down to 63, (from her original 200!). This was extremely exciting, as it showed that the b12 and Betaine were working for her! I ended up diving into some heavy reading that night, to learn more about what Marley's condition.

3/28/22

Step one towards getting Marley home happened that morning. They popped the top off of her incubator, and we were free to pick her up any time we wanted! In order to be discharged from the hospital, she would have to maintain her body temperature and continue gaining weight for a couple of days. The end was starting to be in sight!

HCU HERO: MARLEY FROM TEXAS

3/30/22

Two weeks into our NICU stay, and after a few successful days of weight gain and maintaining her body temperature, we were discharged from the hospital. But first, we were trained on giving Marley daily injections of Hydroxocobalamin, measuring and administering betaine, along with the couple of other liquid medications she was initially put on.

KEY MOMENT

Before we got discharged, one of her NICU doctors came to say her goodbyes and give us her well wishes. She said something that would change the course of Marley's life for the better: "Find a support group on Facebook, because you will not find local support that you will need. Her diagnosis is very rare. Your pediatrician will most likely know nothing about this, and you will need to connect with other families. Search the general terms like 'MMA' or 'HCU' and find support!"

On our way to the HCU Network America Family Conference in June!

That doctor changed everything for us. Before ever walking out of the hospital doors, I got on Facebook and came across a couple of groups; HCU Network America was one of them. Once added to the support group, I made a post introducing our family and sharing the basics. "This is Marlene. She is 4 weeks old, and we think she has Methylmalonic Acidemia and Homocystinuria." (Within the next couple of weeks, genetic testing came back proving that Marley had Cobalamin C, dup 271.A mutation). Many people from the group welcomed us. 4 different moms messaged me asking what dose of Hydroxocobalamin Marley was on. I told them 1 mg/ml. We had no idea that this was considered way underdosed, which led me to ask our new

HCU HERO: MARLEY FROM TEXAS

geneticist about it. I quoted those moms, sharing the information they had given me about high dose hydroxocobalamin, cited an article or two, and then shared the names of a few specialists from the National Institute of Health (NIH), who were willing to talk to our geneticist about the best treatment for CblC. That began my journey of advocating for a high dose of hydroxocobalamin. We are so thankful that our geneticist was willing to talk to Dr. Venditti and Dr. Manoli and their teams.

At a visit with Dr.Venditti

He was on board to increase her dose and learned more about treating her specific mutation of CblC. We are forever thankful, because we know now that time is of the essence, especially how CblC can affect the eyes. The high dose hydroxocobalamin seems to be helping Marley's eyes, and her overall development. She now gets her daily injection of Hydroxocobalamin and Betaine 3 times a day. She weighs about 20 lbs.

My advice from that entire situation is: BE AN ADVOCATE! If you read something, or get a recommendation, it is often worth asking for more information. Don't be scared to question or ask your doctor; a good one will always listen and be willing to explore. While our medications and doses may not all be consistent across the community, there are many families in this journey are willing to share what information they have. If you're reading this, don't hesitate to reach out to me, or any of them!

Our family with Dr.Manoli at the HCU Network America Family Conference

We started educating our immediate family and friends by sharing information. We posted on Facebook, and had countless conversations to explain CblC, what medications Marley needs daily, and how to give them.

HCU HERO: MARLEY FROM TEXAS

We have a document that has everything written out, just in case someone needs to reference it, and we adjust it as her dosages change. I like to call it our cheat sheet, and I've found that the more detail I gave, the less stressful the regimen became. Early on we involved our family members in Marley's care. We had a few of them practice giving her shots after watching us. We felt so much relief that someone else could give her the injection, if for some reason we couldn't.

We enrolled Marley in daycare at 5 months old and began teaching the staff about her medications. While it was hard to let go of control of always having her under our care, it was so freeing when to resume "normal" life and receive help from others.

I will say, the worry never goes away. We worry about how Marley is doing compared to peers. We worry that she will be perceived as "difficult" as she does have more needs than other kids her age. But her teachers are always reassuring us that she is amazing, and not a burden, and it means a lot to us. When you have a child with complex medical needs, you want to protect them in any way you can. We really lean into our faith and try to never project our fears onto our kids.

My husband and I find so much value in learning, advocating, teaching, and just being there for other families in any way we can. This journey is not something anyone should try to navigate alone. We feel strongly that Marley was given her rare disorder for a reason greater than we'll ever understand. God had a plan when he chose her for this and chose our family to navigate it alongside her. As crazy as it is, we are thankful for the growth it has allowed us to have as a family. We appreciate life in a way we didn't know how to before.

HCU HERO: MARLEY FROM TEXAS

While it can be so devastating and difficult at times, know that if you are reading our story and personally navigating the same or similar diagnosis, you are not alone! It does get easier with time. We are a little less than 2 ½ years in, and while it feels like we've learned more information than you should ever learn without a medical degree, the hardest parts of being newly diagnosed have subsided. We love connecting with new families and feel like it is our purpose to serve others, until Marley is old enough to one day connect with you herself!

Our greatest hope is that medication (high dose hydroxocobalamin specifically) can be easily accessible one day. We also hope that families will continue to fundraise for HCU/MMA/Cobalamin Disorders. As a rare disease community, everything we do to raise funds for research will help future generations.

Click here to read Marley'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 VIDEO CONTENT

Our **Moving Mountains Family Conference** in Aurora, Colorado was incredible!

Incase you missed it, or if you just want to enjoy the content again, we've uploaded videos from our conference programming to our YouTube Channel.

Click here to enjoy!

Thank You to our speakers, panelists, workshop facilitators, volunteers, sponsors, exhibitors, and attendees for helping to make this event so impactful.

HCU Champions

INTRODUCING

HCU CHAMPIONS

HCU Champions is a new program designed to help increase awareness and advocacy for homocystinuria (HCU).

HCU Champions will share their journeys and experiences of living with HCU to help inspire, motivate, and empower individuals to connect and support one another in their communities and beyond.

Get involved!

2023 Team features

Team Will for HCU

$2,150 raised for research!

We have run a lot of miles supporting Homocystinuria (HCU) research, but the race is far from won. Rare diseases simply don't get the funding that more widespread diseases do. That's understandable but frustrating. It puts the onus (and running shoes) on the families and friends of those afflicted to fill the funding gap. Please join us in supporting this year's Race for Research to help improve the lives of those afflicted with HCU.

-Chris, dad of Will, who lives w/Classical HCU

Miles for Marley

$2,150 raised for research!

Cobalamin C research has led to treatment changes, even within the last 5 years! Our family is thankful for those ahead of us on this journey and for our village supporting our efforts to learn, advocate, teach others, and help continue funding research for Cobalamin C! In just 2 short years of being on this journey, we've seen our fundraising efforts put in place directly towards CblC research. Our prayer is that Marley, and all families affected, will one day see improved changes in treatment, easier accessibility to their life-saving medication, and standard treatment for all patients diagnosed with her rare disorder.

-Aubrey, mom of Marley, who lives w/Homocystinuria Cobalamin C

https://charity.pledgeit.org/HCURaceforResearch2024

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

Pricing

- **$5 -Participation only!**
- **$20 - Medal or shirt only**
- **$30 - Both!**

Medal deadline is August 8th!

T-Shirt deadline is August 18th!

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

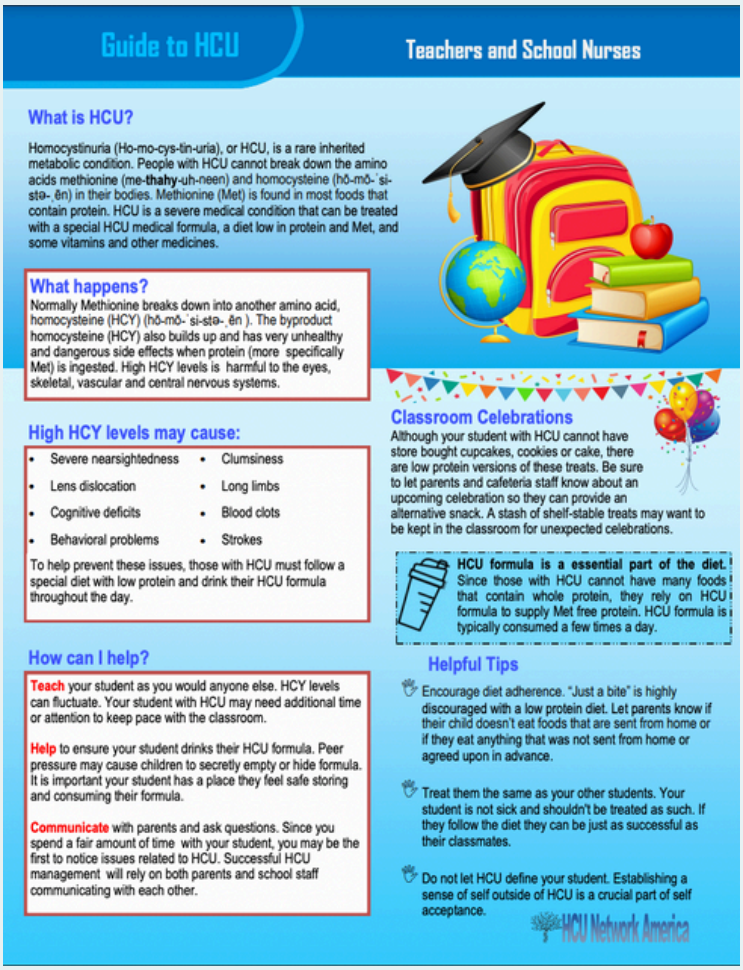
Listen on amazon music

The latest episode

This time, Ben welcomes Brandon to the table!

Brandon's son, Mason, lives with methylmalonic academia w/homocystinuria Cobalamin C. In this episode, we'll hear about how Mason and his family have tackled the challenges presented by vision loss, and how they've turned surviving into thriving!

We have Back to School resources!



Educators Guides

**también disponibles en español*

- Classical HCU
- Cobalamin G & E
- Cobalamin C, D, F, J, X
- Severe MTHFR

Navigating Accomodations

**también disponible en español*

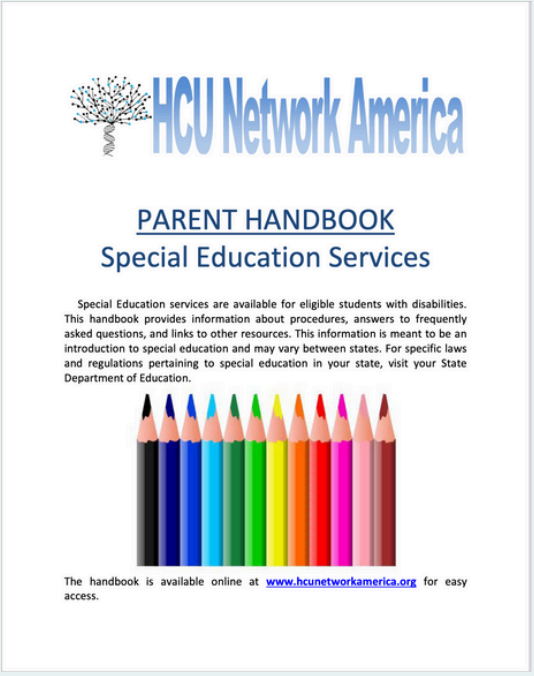
- Parent Handbook for Special Education Services

Low Protein Lunch resources

- Cambrooke school lunch program & more

College Transition

- Off to College Guide



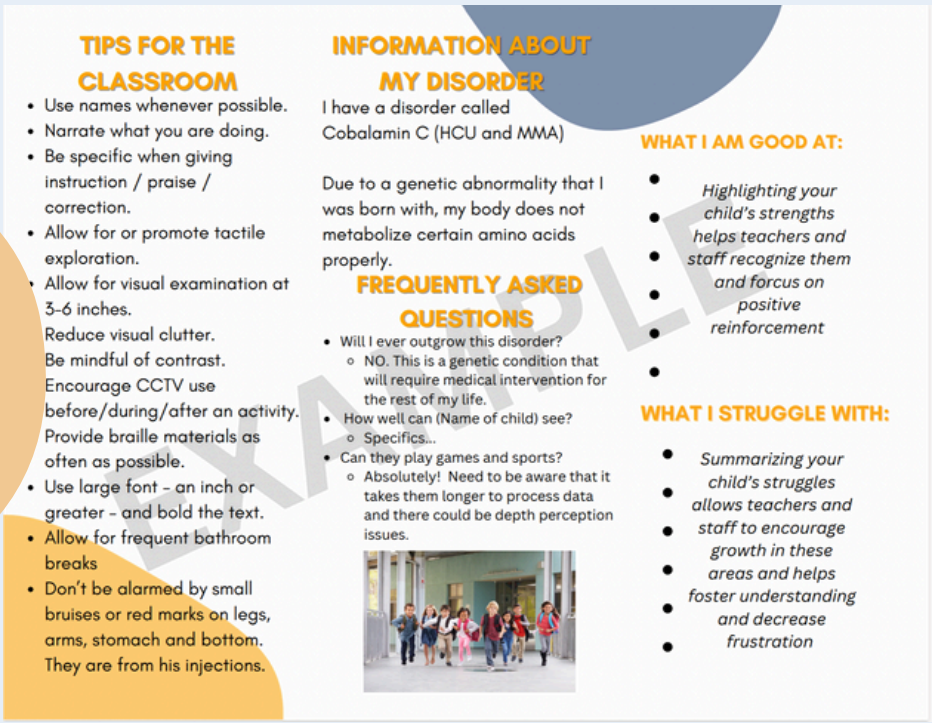
Click here see more Back to School & special edition topics!

We have Back to School resources!

Customizable "All About Me" pamphlet

Designed for those with Cobalamin Disorders or Severe MTHFR

a great tool for teachers/school staff, coaches, babysitters & family members!



Click here to download & get started customizing your pamphlet!

flok is excited to host two in-person family camps this year for our growing community! Our camps are open to all Classical HCU, MSUD, Organic Acidemia, PKU, Tyrosinemia, and Urea Cycle Disorder community members.

Campers are welcome to join us with family, friends, or on their own (18+) for three days of traditional camp activities, crafts, delicious low-protein food, educational sessions, and the opportunity to connect with others in a fun and supportive environment.

East Coast

2024 Dates	Sept 19, 4pm to Sept 22, 12pm
Location	Camp Belknap in Tuftonboro, New Hampshire on the shore of Lake Winnipesaukee
Travel	2-hour drive from Boston, MA 1.5-hour drive from Manchester, NH
Activities & Offerings	Swimming, sailing, archery, ropes course, climbing wall, Science Night, camp Olympics, low-protein cooking, crafts, and social connection
Lodging	Traditional cabins for 10 people; separate bathroom facility
Meals	3 meals/day plus snacks, accommodating all ranges of protein intake & allergies
Accessibility	Natural pathways. Some areas have limited access; golf cart may be available to assist with mobility needs
Medical Accommodations	Onsite New Hampshire-certified medical professional. Huggins Hospital is 7 miles away.
Cost Ages 2 & under are free	$250/camper

Thank to the nearly 300 attendees at flok Family Camp West 2024! We had a great time. Keep an eye on your email for our post-camp survey, camp cookbook, photos, and more! Our inaugural adult retreat was a success as well - THANK YOU to all our participants.

Limited scholarships are available - **click here** to apply.

Volunteer roles and paid work crew positions are available. Contact camp@flok.org with any questions, accommodation requests, or volunteer/work crew interest.

Visit **flok.org/camp** for more details.

July 29, 2024

Subject: Metabolic Supply Update Due To Tornado Damage To Reckitt / Mead Johnson Nutrition Warehouse

Dear Patient / Caregiver,

We would like to report that on Tuesday, July 9, 2024, a tornado struck our warehouse located in Mount Vernon, Indiana. The warehouse sustained significant damage. We are grateful and relieved to confirm that all employees are safe and express our deepest sympathy for those in the community who have been affected by the tornado.

The Mount Vernon warehouse is one of several warehousing facilities we operate across the United States. While the Mount Vernon warehouse is currently out of operation, the rest of our warehouses are all operating normally and we have diverted all inbound deliveries to our other U.S. warehousing facilities.

While recovery efforts are already underway, it is likely over the next month or two that there will be some interruptions to the supply of select formulas, including our metabolic formulas. We are working closely with all our stakeholders including regulatory authorities, medical distributors and suppliers, to minimize disruption, by leveraging our global supply chain and managing inventory at our other North American Nutrition warehouses.

We will provide updates as more information becomes available. If you have any questions, please do not hesitate to contact your local healthcare professional, medical nutrition supplier or our Consumer Resource Center at 1 (800) BABY123.

Thank you for your understanding in the short term as we work through this natural disaster.

Warm regards,

Cindy Hasseberg, Ph.D., R.D.
Vice President of Medical Sales
Mead Johnson Nutrition

Morris Corporate Center IV
399 Interpace Parkway
Parsippany, NJ 07054-0225

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

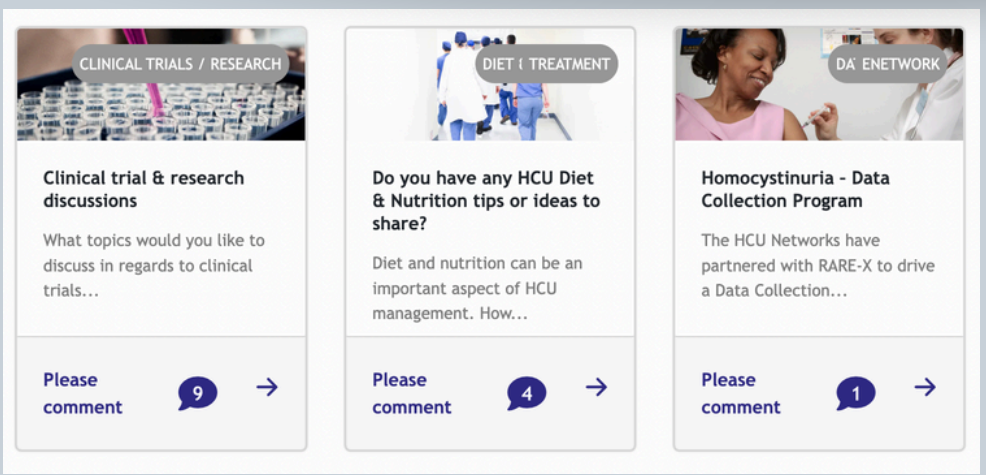
TRAVERE THERAPEUTICS

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/

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

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

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

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FOLLOW US