The HCU Herald

Featuring...

HCU Hero
Trace from NC

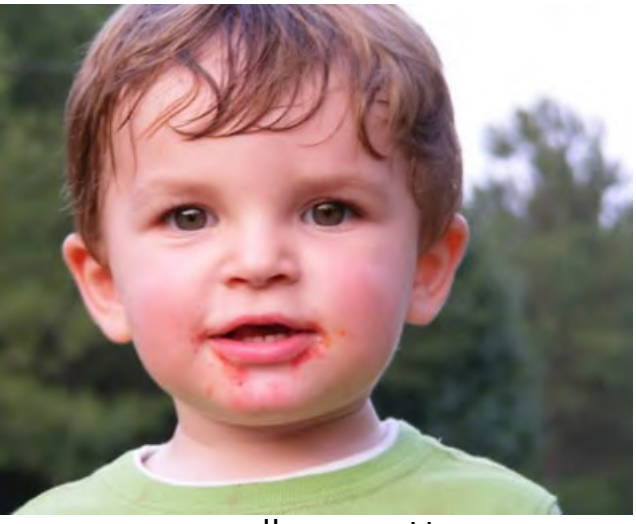
March 2025

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

HCU HERO: TRACE FROM NORTH CAROLINA

In August of 2005, I got some of the best news of my life: a positive pregnancy test. My husband and I struggled for about two years to get pregnant and ultimately required help from the doctor with medication. My husband and I were excited and declared this pregnancy a true miracle. My pregnancy was uneventful, other than having gestational diabetes, though at the time I was working as a NICU nurse and was anxious about any and everything that could go wrong.

On May 23rd, I went for my 39-week checkup. After a non-stress test, my doctor wanted me to go straight to the hospital for induction. My labor was uneventful for the most part, until pushing. Trace was choosing to stay put. The vacuum extractor had to be used and when his head was finally delivered, we found out why: he had the umbilical cord tightly wrapped around his neck. When he was fully delivered, the doctor showed him quickly to me and then he was whisked off to the nurses. At the time it didn't occur to me, but looking back he was not a pretty pink; in fact, his color was a bit bluish. I think he cried but I am not really sure. He was given to me not too long after and our stay at the hospital was pretty uneventful. He was a bit jaundiced, which was monitored, but great otherwise. As far as his newborn screen, I was never notified of any issues.



Trace was the perfect baby and toddler. He met every milestone, some a little on the late side, but still on time. He never cried, except when he was hungry. As a toddler we did notice that Trace would flap his arms when he got very excited. We thought nothing of it; just thought it was a quirk of Trace. It would become significant later, though, when he began school.

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After starting school, Trace's teachers expressed many concerns over his behavior, like flapping his arms. Finally, toward the end of third grade, he was diagnosed with Autism and ADHD. This diagnosis seemed out of the blue for my husband and me and at that time we began to mourn the future that we had envisioned for Trace. We soon found out that my husband's brother (who is 14 years younger than he) had been diagnosed with Asperger's and that actually gave me hope for Trace's future. I knew that my husband and I would do everything we could to help Trace be successful in life. He had an IEP at school, and we began ABA therapy. All of that gave us hope for Trace's future independence, though we still didn't know what his future would look like. Every year we saw him mature, do better at school, and just become a wonderful person. In fact, in seventh grade, we terminated his IEP at school because he had done so well that year.

Then came the 2019-2020 school year, eighth grade. Everything seemed great at the beginning. Then he started vomiting. I would get phone calls from school often of him vomiting. At the beginning, we thought it was constipation, an issue he has had throughout his life. His pediatrician decided to admit him into the local hospital to have treatment for constipation. He tolerated that stay pretty good, though we did find out there that he is a hard stick. We left the hospital hoping that this was going to fix the problem, but boy were we wrong. He continued to have episodes of vomiting, mostly at school. I am grateful for my parents that picked him up from school, because then I was teaching at a high school and couldn't always leave.

Trace ended up being referred to gastroenterology at UNC (University of North Carolina, Chapel Hill) and hospitalized twice for vomiting. They did every GI test

they could think of. He had a colonoscopy, endoscopy, swallow studies, CT scans and MRIs. Everything came back normal, and his dad and I were so frustrated. Of course wanted nothing to be wrong, but at the same time wanted to find an answer that would explain the issues. Finally, after a psychologist spoke with Trace, we got what we thought was the answer. Trace said that he was being bullied at school; something he had never mentioned to us before. After that, they diagnosed him with Functional vomiting, which basically means that there is no physical reason for the vomiting but that was probably psychological. His father and I were in agreement with this diagnosis. Trace has never been very good at telling us what is going on with his body and we knew he had anxiety issues that manifested as nausea. After this diagnosis, we changed his school schedule, and everything seemed to even out, except his eating. When he started vomiting, would quit eating altogether. He basically survived on Ensure, having to drink six every day. During this time he lost an extreme amount of weight. We noticed a couple of times that he seemed weak, but thought it was because of him not eating and due to the weight loss.

Then in late November, he began falling for no reason. I knew he was falling, but again thought it had to do with his eating habits or not eating. I also wasn't aware of the extent to which he was falling until his grandmother pointed out that he was falling a lot. It wasn't that we hadn't noticed, we just wanted some normalcy from the months before and didn't want to think something else could be wrong. I even called and talked to my mother about how clumsy I was as a teenager. She agreed that I was clumsy, but pointed out that I didn't fall a lot. After that, I started thinking that maybe something was really wrong. Then, one of his teachers called and said he had had a few falls at school but would tell them that he had just tripped. His teacher said that day she saw him going down the hall with nothing around and witnessed him just fall, like his legs just went out. I made an appointment with his pediatrician, which happened to be a doctor that I had worked with at the local hospital. I was hoping it was a Vitamin B12 issue that

could be solved by a simple injection. Boy, was I wrong! The pediatrician could not elicit the knee jerk reflex, which made her very concerned. She suggested I take him to a children's emergency room. I called his dad, and he met us at UNC. That began the journey that led to the final diagnosis.

At the children's ED it truly felt like the attending neurologist read his chart and previous diagnosis and decided his diagnosis before she even saw him. She told us that he had "Functional Falling" that it was all in his head. I pushed back, explaining that there was something very wrong with my child, and it was not in his head. The MRI we did the night before didn't show anything, so she didn't think anything was physically wrong with him, although she never watched him walk. The physical therapists that brought him his walker mentioned to us that his walk was off. The neurologist agreed to refer us to physical therapy and follow up in two weeks, except when I got the call to make the appointment, they said the earliest they could see him was in six weeks. I was frustrated and furious. It felt like no one was really listening that something was really wrong with my son. My son was 13, 5 foot 8 inches and weighed over 200lbs. How was I to pick him up every time he fell? He could not get up by himself. I was at a loss for what I needed to do. Trace's grandmother encouraged me to try and find a neurologist that we could get in with quicker. I called around and found a neurologist that could see him.

As soon as we met this new doctor, I had so much hope. She did a very detailed assessment, watched him walk and agreed that something was very wrong. She decided to draw a bunch of labs and to do genetic testing. It felt like finally we were going to figure out what was going on and that there was someone that would help us fight for an answer. Many potential diagnoses were mentioned. I also took to Google, trying to figure out ways to help. Having a medical background is good when things like this happen but can also be very bad. For a short time, I thought he could have ALS, which has a poor prognosis. My husband and I tried to keep a brave face for Trace. I truly struggled. I knew that the only thing I could do was to trust God to keep us and Trace in his hands. I knew

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God had a plan for Trace and would keep him. During all this, Trace never asked why God allowed this to happen. He always had the best outlook. He knew God had him and that we would find out what was wrong.

The labs drawn by the neurologist came back pretty much normal. I of course looked up every lab and what it could mean. I noticed that one lab had a very abnormal value. It was a urine acid organics - methylmalonic acid. It was extremely high. I called the neurologists about it and she said she had already referred Trace to UNC genetics, but would call and see if she could expedite the appointment. Though I was still upset over the events at the UNC ED, I agreed to the referral because I trusted the neurologist to send us to the best doctor for Trace.

Our first appointment with the geneticist went really well. A lot of the genetic tests had came back. We discovered that Trace is a carrier for Pompe disease. We were told, though, that it usually doesn't affect the carrier, but we would watch it. There was also a variant gene for a type of muscular dystrophy. I asked the geneticist about the elevated levels of methylmalonic acid that had showed up in Trace's labwork. She said she really didn't think it was a metabolic disorder, but would draw additional labs to check. I will never forget the day I got his diagnosis. I was at a new teacher training session and had to step out to join a phone call for an appointment for the neurologist that my husband had taken Trace to. I was trying to be as present for the training as I could be, but it was hard. During a break, I checked my phone and noticed I had a message from the geneticist that we had a diagnosis. I ran back to the training room, grabbed a pen and paper and ran back out. I later apologized to the trainer. I was so excited. We finally had an answer.

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His geneticist told me he had MMA (methylmalonic acidemia) with homocystinuria. He started on daily hydroxocobalamin injections (we started betaine sometime later). I started trying to read everything I could find, and it was a little confusing for me at first. Everything I could find was about infants with MMA. I also didn't know the type yet. The first appointment with his doctor after the diagnosis was amazing. She talked to us about the diagnosis, the treatment and the hopeful prognosis. She told us that she had other patients with MMA, but Trace was her first with late onset. She was very hopeful that he would get full use of his legs back and that we could control symptoms and progression through medication. During that first appointment, Trace's doctor was going over the usual symptoms. My favorite thing was as she was going through the symptoms, she got growth retardation. She looks at Trace's 5'9", 250 lb self and says, "I don't think that is a problem." She wrote out about the metabolic cycle on the paper on the table, which I still have today. She gave me many different articles about MMA as well and suggested that I find support groups online and on Facebook. Going home after that appointment felt a bit overwhelming. There was so much information, and even as a nurse I had a hard time understanding it all. To tell the truth, I still don't understand the whole metabolic pathway and how it all works.

Coming home after the appointment and telling all our friends and family was a little difficult. All the information I had was about children that had been diagnosed in infancy. I had to remind them that Trace was different, and that a lot of the symptoms and some of the complications didn't really apply to him. Our family and

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our church were very supportive, but this was all happening in early 2020, when everything was locked down. Being a teacher who was now working from home meant I got to spend more time with Trace, which gave me the opportunity to try to digest how our life was going to look. But, boy did we have to adjust to our "new normal". Trace was using a walker but could still go up the steps of the porch. We had to get a shower chair for the shower because he could no longer stand for long periods. We borrowed a wheelchair from a family member, so he could go do simple things with us, like going to Wal-Mart. He had to learn how to dress, because he could no longer stand on one leg. We had to buy a contraption that had poles to help him stand from our couch. I think his dad and I had a harder time with all the changes than Trace did. He always took everything in stride and told us often that God had his back.

Trace went to physical therapy for approximately two years. He started out at three days a week, then two, then one and finally discharged when we felt like it was as good as it was going to get. He went from a walker to two forearm crutches to one. He struggled with his confidence to change until he was pushed to use them consistently. We realized after about a year and a half that he was never going to get full use again but decided that we were okay with how far he had come. His doctor still believed he was going to get better, but we didn't want to keep pushing and prepare for him to never get use completely back. We also found out that damage had been done that had caused his legs to buckle whenever he received a fright, and that this caused him to fall many times, and ultimately led to him falling about a year ago and having to have surgery on his ankle. That surgery was his first major event since

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getting sick and getting the diagnosis. His dad and I were so nervous about the surgery. We contacted the geneticist's office, but our doctor was out of the country. The doctor on call was awesome and called the anesthesiologist to discuss his care. We found out on the day of surgery that the anesthesiologist's daughter also had a metabolic disorder, so he completely understood the importance of Trace's issues. Knowing this put my husband and me at complete ease. We trusted him completely and Trace came through surgery with flying colors. After the surgery, he used his wheelchair a lot more before learning how to walk again, first with a walker, then with crutches.

Trace has overcome so much throughout the last five years and he is doing so good now. He graduated from high school last year and is currently going to a community college. He is working on getting his driver's license, using hand controls. My hope for him is for him to be independent. We know that he may need us close to help with certain things but fully believe that he will be independent one day. He currently does most of his medicine himself. He can do his shots but mainly likes his father (not nurse mother) to give him the shots. His dad currently sets up his meds for the week, but we will start getting Trace used to doing that as well.

I never thought this was going to be our path. When Trace was diagnosed with autism, I didn't know what his life was going to be like. I started to feel good about his future when he was in seventh grade. In eighth grade, I was hit again with an unknown future, and I still don't know what the future will hold. However, I know that Trace will meet it with his usual gusto and determination with his family and God by his side.

Click here to read Trace's and other patient stories on our webiste!

NEWLY TRANSLATED RESOURCES

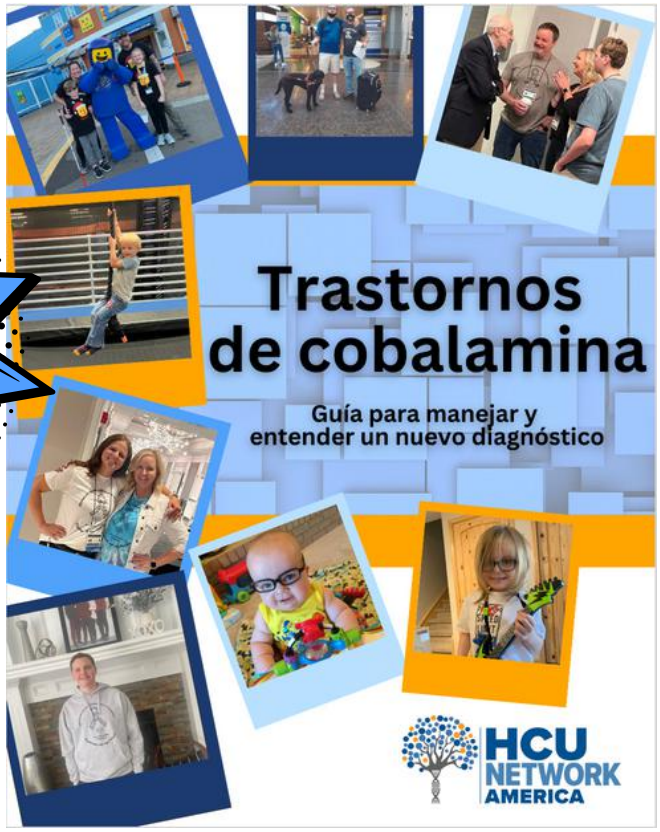
Classical HCU Toolkit

Now in Arabic!

DOWNLOAD NOW

Cobalamin Disorders Toolkit

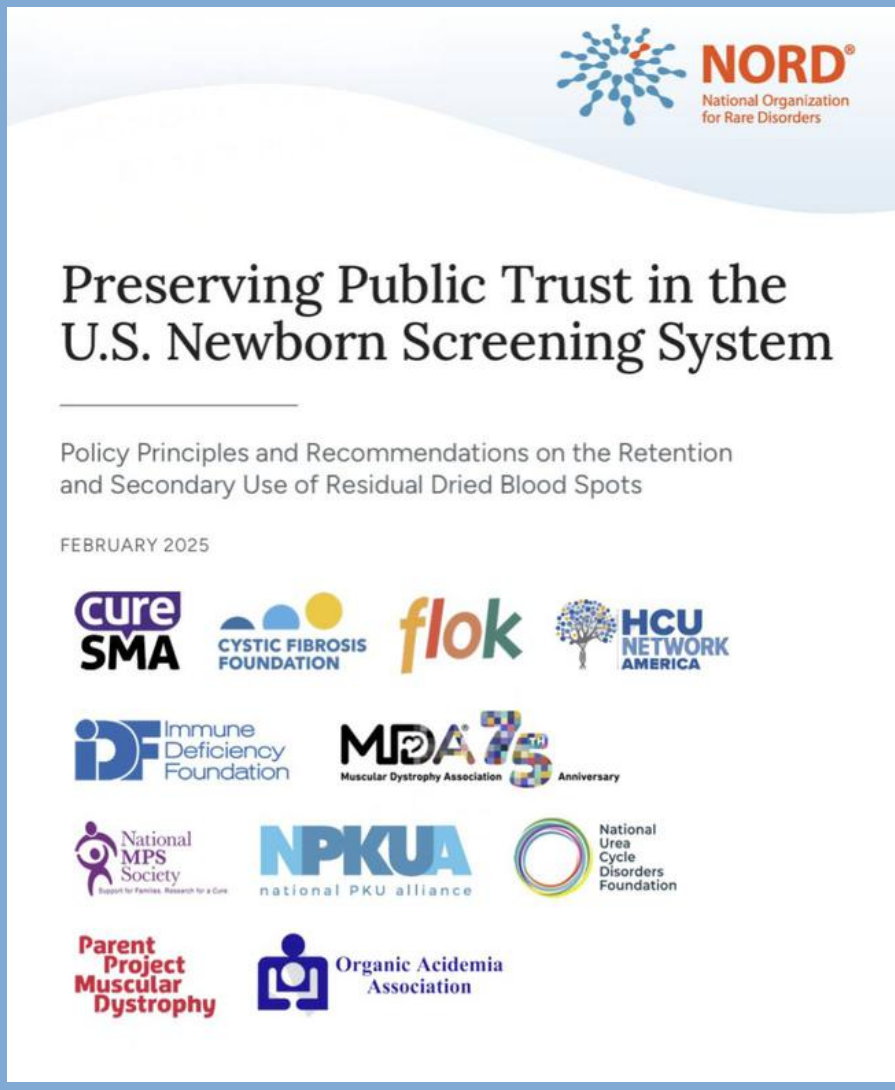
Now in Spanish!



DOWNLOAD NOW

NEWBORN SCREENING

Classical homocystinuria is **screened for in all 50 states but is missed approximately 50% of the time**. Cobalamin disorders and severe MTHFR are also screened for in some states but are often missed.



Over the last six years, HCU Network America has partnered with state newborn screening labs, the CDC, and other organizations to improve newborn screening methods and help ensure all babies born with homocystinuria are diagnosed on time and able to start treatment immediately.

We are proud to share our involvement in a new paper from the **National Organization for Rare Disorders (NORD®)**, outlining policy principles and recommendations to fortify public trust in the U.S. newborn screening system.

Learn more:
https://rarediseases.org/newborn-screening-report/

FOOTBALL SQUARES FUNDRAISER

RESULTS

$900 RAISED, TO BENEFIT HCU NETWORK AMERICA'S EDUCATION & OUTREACH PROGRAMS!

CONGRATS TO OUR WINNERS!

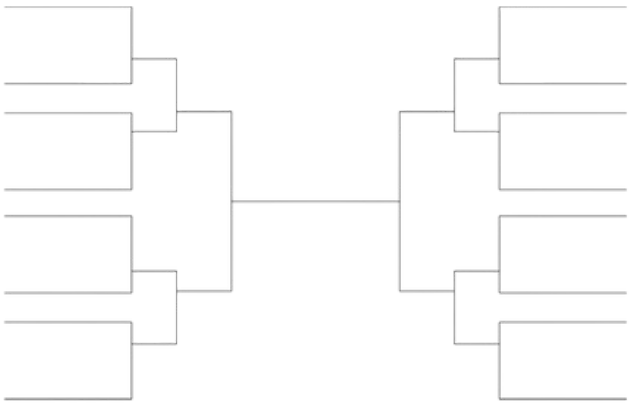
- THE HUMMEL FAMILY
- ROBBIE H
- SHANE P
- AUNT MAGGIE S
- AMY N
- JACQUELINE P
- KRISITN R

THANK YOU TO ALL WHO PLAYED!

HOST YOUR OWN MARCH MADNESS FUNDRAISER!

March Madness Selection *Sunday 2025 is* ***March 16!***
Start assembling your pool & raise funding for resources, education & outreach for **HCU Network America!**

March Madness is a three week period packed full of buzzer beaters and is a sports poolers dream. But how can you keep the excitement alive when most of your members have had their brackets busted? A great option is our Madness Squares pool format, as every game of the tournament will have a winner!



How Do Madness Squares Work?
If you are familiar with Super Bowl Squares, the main idea is the same for March Madness. A 10x10 grid of boxes is setup and each row and column is given a number from 0 to 9. Just like in Super Bowl Squares, each square of the grid can be claimed by a pool member.

Winner breakdown
Each round is worth a set number of points. You can determine this on your own, but be sure to let all of the entries know before the tournament begins what the scoring system will be. (You should write the point values under each round at the top of the bracket).

Declaring a Winner
Multiply the total number of correctly picked games in each round by the points assigned to that particular round. Tally all rounds together and the person with the highest point total wins!

- For further instructions and to print your bracket, visit: https://www.printyourbrackets.com/howtomarchmadness.html
- For online tools, check out: https://www.runyourpool.com/march-madness-squares-pools.cfm

UPCOMING EVENTS

Happening Sunday, 3/9!
Register now!

HC&U Podcast

Meet your hosts!

Welcome to the HC&U Podcast!

We're Ben and Lindsey, your hosts. We are so excited to host this podcast as extra resource for the Homocystinuria community. We hope you like our content!

Read Ben's patient story here.

The latest episode

Ben welcomes Trace & Jessie to the table!

Jessie is the mother of our March HCU Hero Trace, who was diagnosed with late-onset Cobalamin C. Jessie, Ben & Trace will chat about the brain and nervous system, how Trace's symptoms presented, and their journey to diagnosis. Trace will discuss his experiences & what he's up to now!

To Listen:

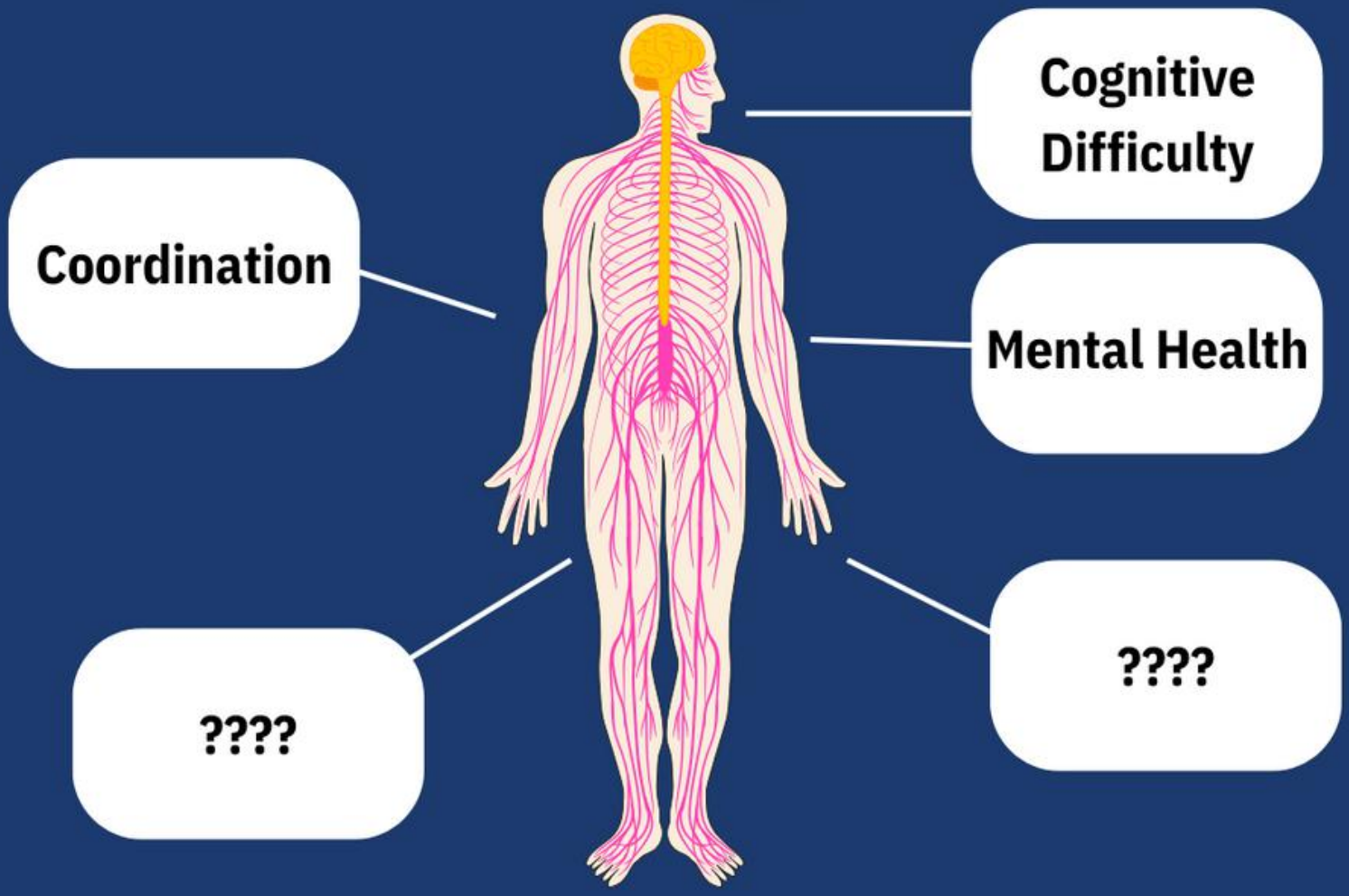
https://hcunetworkamerica.org/hcu-podcast/ or click your favorite option below!

LISTEN ON Spotify | LISTEN ON iHeartRADIO | Listen on Apple Podcasts | Listen on amazon music

HOMOCYSTINURIAS DATA COLLECTION PROGRAM

POWERED BY RARE X

Help Fill In The Gaps: The Brain and Nervous System



Take the Brain & Nervous System Survey:
https://rare-x.org/homocystinuria/

CANPCU+ CANPKU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

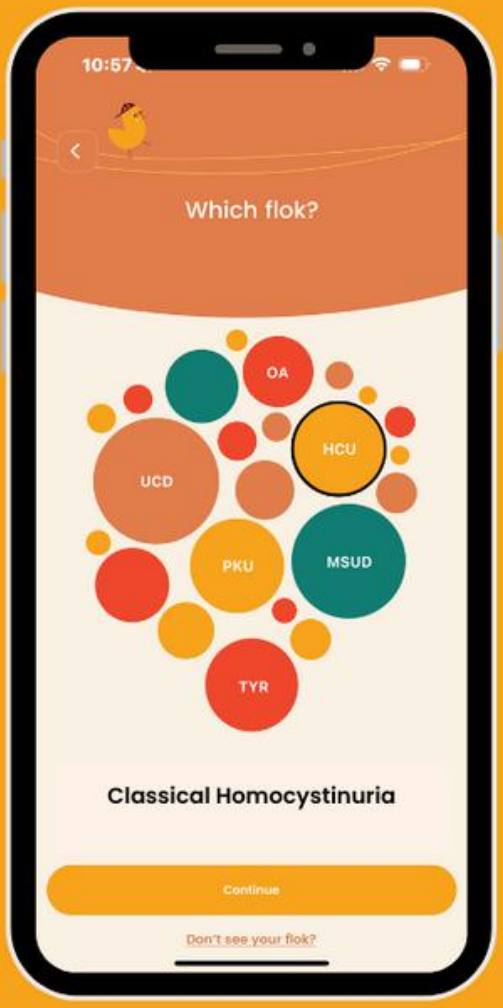
Start using the flok app today!

Welcome to the next generation of metabolic care for the CLASSICAL HCU COMMUNITY

The flok app is now in Open Beta in the United States.

Download at flok.org/app

flok



Big news!

The flok app is now in Open Beta, available for download to everyone in the U.S.!

Jump in and start exploring - the app helps you manage your diet and build a full picture of your metabolic health - including symptoms & moods, activity, medications, and lab tests.

Scholarship opportunity!

flok The Guthrie-Koch Scholarship

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

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

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

Click here to learn more and to start your application!

The application deadline is March 15, 2025.

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*!

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