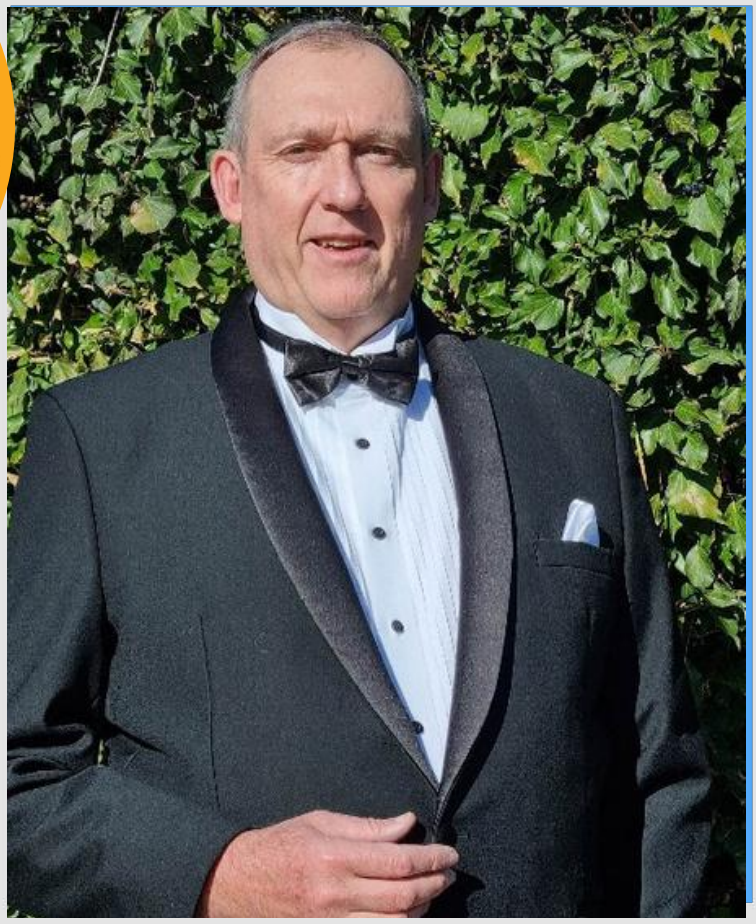
The HCU Herald

World **H**omocystinurias **A**wareness **D**ay Edition!

Featuring...



HCU Hero
Michael from Australia

May 2025

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

HCU HERO: MICHAEL FROM AUSTRALIA

My name is Michael Stevenson. I have outlived both my parents and all the clinicians who were involved in my early diagnosis and treatment, so what follows are my memories as I was told and as I remember them.

Twins Michael & Phillip, 1961

I was born in Sydney, Australia in 1961, and I was not alone. This was my mother's third pregnancy, with an older brother Darryl born in 1956, and an older sister Kerri, born in 1958. Carrying twins, my mother was preparing to go into confinement for the last month when she went into premature labour. We had not yet turned so we were born a month premature, and we were born breach. It was not an easy birth. I was born first, my brother Phillip arriving after me, with my umbilical cord wrapped tightly around his throat. Within hours of our birth, Phillip began to fit, and he was placed in intensive care, not expected to live. It would be nine days before we were allowed home.

These days, Phillip would have been diagnosed as suffering acute cerebral palsy. The diagnosis at the time was severe mental retardation and spastic paraplegia. Phillip would never learn to talk and was only just learning to walk at age 11 when he passed away.

By the age of 4 he became too much to handle for my mother with three other healthy children and he was placed into care during the week, initially coming home on weekends, but eventually moving into care full time. It was whilst in care that Phillip underwent emergency surgery for an acute urinary infection which entailed the removal of a kidney stone that was described as being "the size of a duck's egg", around 1966 - 1967.

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One of the doctors at that hospital was Dr. Brian Turner who specialised in psychiatric medicine. He noticed the blond hair, long limbs, and high arched palate associated with Homocystinuria, a disorder that had only been recognised less than five years before, with only 29 known cases, all of them in Ireland. Testing Phillip proved that he had indeed found the first case outside that country.

Michael, Phillip, and mother, 1962

A 1964 paper entitled "Pathological findings in homocystinuria" by J.B. Gibson, Nina A.J. Carson, and D.W. Neill from Belfast noticed the similarities to Marfan's Syndrome but stated: "Mental defect appears to be an inevitable element of homocystinuria, for the condition has been detected only in mentally retarded individuals." Indeed, it was thought that as Marfan's Syndrome was known not to cause mental defect, that this was the defining point of difference between the two disorders. You can see that paper here.

My siblings were also tested, as were my parents. My older brother Darryl showed no signs of the disorder, but my older sister Kerri was also positively diagnosed. Hers turned out to be slightly more severe than mine with the addition of a sunken chest and a possible cerebral thrombosis soon after diagnosis. She was also exceptionally tall for her age and at 13 was put onto hormones to stop her growing. This led to a lifetime of problems. She passed away at the age of 65 from cancer.

However, Dr. Turner's discovery of Australia's first HCU patient came with a bombshell. Not only was the patient an identical twin, but that twin was certainly not intellectually disabled, disproving the previous hypothesis that HCU inevitably led to intellectual defect.

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3-year-old Michael, 1964

I also have the long limbs, pes cavus, and high arched palate, and like my sister, was initially blonde. Growing up, we were both exceptionally skinny and could not put on weight. At the age of 25, I was 6'5" and weighed less than 160 lbs. Interestingly, when having root canal treatment, I was told that my root canals are about a quarter of an inch longer than normal, so I guess it is not only the skeleton that is long and lanky.

As the first "non-retarded" individual ever diagnosed with HCU, I became a guinea pig and was subjected to test after test. I can still recall as a six-year-old having electrodes strapped to my chest and my head to measure both my heart and brain, looking with interest at the lines on the screen whilst wearing a net on my head for the EEG, and being asked all manner of questions, as well as sitting at a desk in a corridor doing a series of IQ tests. No previous patient had exhibited an IQ higher than about 60. At the age of six, they were unable to measure my upper limit and marked me down as 130+. I have always considered my sister to be more intelligent than I am and in a 1978 paper, we were singled out as: "a boy and girl of superior intelligence."

My celebrity as the first patient ever diagnosed as having a "mild" form of the disorder, brought Victor McKusick out from the United States to see me. I can recall him as a tall figure standing over me in a lab coat, asking me questions. I recently discovered that he too was an identical twin.

He asked me if there were any other symptoms I thought were relevant and I tried to describe an uncomfortable feeling in my legs, like butterflies in my stomach that made me constantly twitch my legs up and down to try and get some relief. He dismissed it as nothing but years later it would be officially designated "restless legs syndrome" and recognised as one of the symptoms of HCU. The feeling eventually faded with treatment but still occasionally returns if I sit still at a desk for too long.

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Little was known about HCU at the time. We both had weekly blood draws where Dr. Turner would arrive in his MGB sports car to take our blood then race across town to get it tested in time. This was not a process I enjoyed, and I would frequently make myself scarce before his arrival. On one occasion, my uncle who was a police motorcyclist turned up in full uniform and when I excitedly went out to greet him, he grabbed me in a bear hug just as Dr.Turner's little sports car rounded the corner. I always fainted following blood being drawn and continued to do so until well into my 20's.

There were no supports available as there was no one else with the disorder. The first treatment tried was Pyridoxine Hydrochloride in suspension. This was described as a "cherry flavoured" liquid and to this day I still gag at the memory of its foul taste. It was only available on prescription. Of course, now it is known as Vitamin B6, but this was in the days before dietary supplements were really a thing. Eventually, it became available in tablet form. Unfortunately, it only came in 25mg tablets, and I needed 250mg, so my mother and I would sit at the table filling gelatin capsules with 10 tablets each so that I could take them all in one go. Eventually a 5mg tablet of Folic Acid was added to combat anaemia.

Soon after beginning treatment, both my sister's hair, and my own darkened. Phillip's however, remained blonde. It was only recently that I discovered why. In 1973, he became severely anaemic, slipped into a coma and passed away. We were 11.

Despite attending five different Primary (Elementary) Schools, I managed to be the dux of my last one the year it opened. I went on to be classed in the top 20% of my state in my final exams and went on to university. I eventually served with the Australian Federal Police as a member of the Bomb Squad, as well as fronting their media unit and operating their puppet mascot, Constable Kenny Koala.

Michael and his sister Kerri at AFP Graduation, 1986

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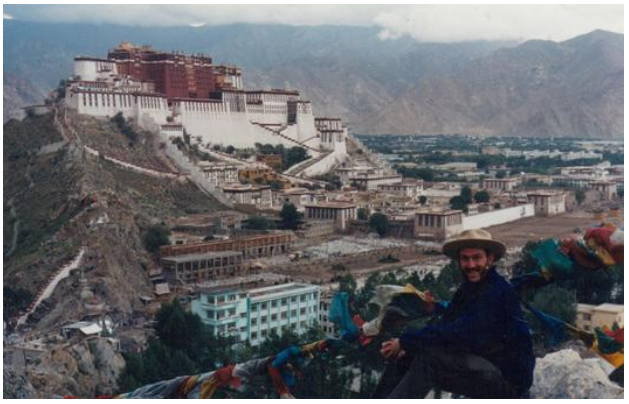
Michael with Mascot, Constable Kenny Koala, 1991

My sister Kerri became a registered nurse, specialising in mental retardation, and eventually worked at the same hospital where our brother died. At one lecture during her training, HCU was mentioned with one of its defining symptoms being mental retardation. Needless to say, her hand went up and she had something to say about that. It took many years for the broader medical profession to catch up.

Initially, the mention of HCU in a hospital setting would bring doctors running to see me but this no longer elicits such a response. However, I still get junior doctors coming to check out my palate, so I guess that is still the stand-out feature.

The only way HCU really affects me is that I am put on blood thinners whenever undergoing surgical procedures in case of blood clots. I have had all the saphenous veins stripped from my legs but have not had problems with my eyesight or cardiovascular issues (apart from the collapsed veins). I have recently added Vitamin D to my daily intake as I have mild osteoporosis in my lower spine on top of slight scoliosis as I didn't stop growing until I reached 6'5".

In Salzburg, Austria, 1995



In Lahsa, Tibet, 1995

I have travelled much of the world, and whenever I travel, I carry a doctor's certificate stating the medications that I require but luckily, the majority are freely available dietary supplements and so I have never had an issue.

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I rarely eat red meat anyway but when overseas I become a vegetarian, mainly because if you are going to get ill it will be from tainted meat or seafood. I am extremely lucky that I have never really had to be over-vigilant about my protein intake although I do avoid anything that announces that it is high in protein, or "a good source of protein" and I am sure that both Kerri and I were a nightmare for my mother growing up, balancing the low protein needs of her two youngest children, with the high protein needs of her eldest.

In 1985 we both took part in a trial to see if Betaine was beneficial to B6 responsive patients. In our case it was not, so my treatment continues to be ingestion of B6 and Folate, with B12 also being added relatively early on. My levels are fairly stable at between 55 and 75.

As a child my mother told me that I was a very fussy eater, living primarily on Vegemite sandwiches. I still eat it several times a week. Vegemite is high in B vitamins, so it was my body telling me that I needed this stuff.

I first took out a Life Insurance Policy in my 20s and was only covered until I turned 55 as I was not expected to live longer than that. I have recently taken out a new policy without pre-conditions, as it is now known that you can have HCU and still lead a long and active life.

Today I have a very healthy daughter, Mycle, and a beautiful granddaughter, Gabby.

Michael with his daughter, Mycle

Michael's granddaughter, Gabby

HCU HERO: MICHAEL FROM AUSTRALIA

Phillip was the 30th person to be diagnosed worldwide with HCU. Every single one of those 30 people were intellectually disabled. The 1964 paper cited above came about through a "systematic search for metabolic abnormalities in mentally retarded individuals in Northern Ireland." Two years later, a search involving the testing of 1500 institutionalised mentally retarded patients was conducted in New South Wales, without finding a single case. This was not long before we moved from Victoria, to live in New South Wales.

When I last spoke to my geneticist (I have outlived my initial treating specialists, who all worked out of psychiatric units) I broached the subject of longevity. All 30 people diagnosed before me (including my brother Phillip) were intellectually disabled. It is a sad fact that people with such disabilities generally have shortened life spans. It is almost certain that all 30 of those patients have since passed away, meaning that although I may not be the oldest person diagnosed with HCU, at 63 years of age, I have probably lived with a diagnosis longer than anyone else alive.

It is only recently that I discovered a paper written in 1973 by Drs Bridget Wilcken and Gillian Turner that stated that treatment was deliberately withdrawn from my brother Phillip. As the first set of identical twins discovered, and the first to be diagnosed with a mild form of the disorder, we were being used as an experiment. I was being treated whilst Phillip, who was uncommunicative, institutionalised, and apparently expendable, was being used as the control. His death certificate states that he died from severe anaemia and heart problems.

"As the first set of identical twins discovered, and the first to be diagnosed with a mild form of the disorder, we were being used as an experiment. I was being treated whilst Phillip, who was uncommunicative, institutionalised, and apparently expendable, was being used as the control."

Without Phillip's chance diagnosis, and the demonstrable fact that his mental retardation was not caused by HCU, as embodied in me, his non-retarded identical twin, how long

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would it have taken for the search to widen outside the institutions for the mentally disabled? How many lives were improved by being diagnosed, where a diagnosis would not have otherwise been considered or looked for?

It astounds me how far we have come in the diagnosis and treatment of HCU in my lifetime, from only having one known form of the disorder to the kaleidoscope of variations now described, with people now discussing which of their genes are affected. (I still don't know mine).

I strongly believe that my brother Phillip should be honoured for his role in the detection and diagnosis of HCU and the consequent realisation that it was a much more diverse disorder than originally imagined. He paid for that knowledge with his life.

In loving memory of Phillip, and with deep gratitude for his role in the development of a deeper understanding of HCU.

SAVE THE DATE

Syntis Bio

18 MAY
WORLD HOMOCYSTINURIA AWARENESS DAY

RAFFLE & PANEL LIVESTREAM

HOPE CONNECTS US AROUND THE WORLD

WE WILL MATCH $10K!

FOLLOW TO RECEIVE UPDATES ON ACTIVITIES AND EVENTS

Host: Ben Massengale
Living with Classical HCU
Georgia

Brittany Hunter
Caregiver, Classical HCU
Mississippi

WORLD HOMOCYSTINURIAS AWARENESS DAY PANELISTS!

TUNE IN ON
MAY 18TH AT 4:00PM EST

Click here to Register!

Brandon Tornes
Caregiver, Cobalamin C
Texas

HCU NETWORK AMERICA

Syntis Bio

Erica Arnaud
Family Member,
Severe MTHFR
Louisianna

HCU NETWORK AMERICA

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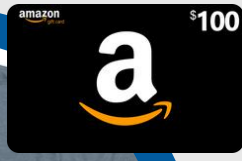
POSITIVE SUPPORT GARDEN

SIGNED SPORTS BASKET

WORLD HOMOCYSTINURIAS AWARENESS DAY

KENDRA SCOTT JEWELRY & BLANKET BY DIANE



HCUNA BASKET

RAFFLE BASKETS

OPEN MAY 5TH-MAY18TH

CANE'S BASKET

CLICK HERE FOR THE RAFFLE!

GREAT WOLF LODGE

FAMILY VACATION BASKET

WORLD HOMOCYSTINURIAS AWARENESS DAY

50 / 50 RAFFLE

Half of the total money raised is awarded as a cash prize to the winning ticket holder, with the other half going to HCU Network America.

1 Entries $5 +

5 Entries $20 +

Click to purchase entries & win big!

HCU NETWORK AMERICA

Syntis Bio

Shine Bright, Do Good

JOIN US FOR A
KENDRA GIVES BACK EVENT

DATE & TIME

May 4th 1pm-4pm

DETAILS

Shop Instore at Kendra Scott Grand Blvd for 20% of your purchase to Giveback to **HCU Network America** on **Sunday,5/4** Can't make it? Shop online at Kendrascott.com and enter code **GIVEBACK-IXPYA** at checkout to give back or call our store at **(850) 240-5270** to place a phone order for free shipping from **5/4-5/5.**

KENDRA SCOTT

HCU NETWORK AMERICA

RECENT EVENTS

American College of Medical Genetics (ACMG) Annual Clinical Genetics Meeting

Los Angeles, California

March 18-22, 2025

American Academy of Neurology (AAN) Annual Meeting

San Diego, California

April 5-9, 2025

While in California, we had the opportunity to meet up with two our of incredible HCU families! The family of Vasco, who lives with Cobalamin C, as well as the family of Luciano, who lives with Cobalamin G.

RECENT NEWS

At HCU Network America, we advocate every day for not only the Homocystinurias but rare diseases as a whole. Whether it be Classical HCU, Cobalamin Disorder, or Severe MTHFR, each of these conditions relies on early detection to access lifesaving care and avoid life-threatening side effects.

We are deeply concerned by the sudden termination of the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC)—a vital source of expert guidance that has shaped newborn screening policies for nearly two decades.

The ACHDNC was pivotal in recommending which serious, treatable conditions should be included on the Recommended Uniform Screening Panel (RUSP)—a tool that helps states decide what diseases to screen for at birth. Its removal leaves newborn screening systems without clear, science-driven oversight, putting countless children at risk of delayed diagnosis, missed treatment opportunities, and preventable suffering.

Each year, newborn screening helps identify approximately 14,000 babies with serious conditions. Eliminating the ACHDNC jeopardizes those lives—and creates devastating uncertainty for families already in the process of advocating for their child's rare condition to be added to the RUSP.
We remain committed to protecting and strengthening newborn screening programs nationwide. But we can't do it alone.

Take action today:

1. **Call your Senators & House Reps** and urge them to prioritize the reinstatement of the ACHDNC and support robust, evidence-based newborn screening.

Your voice can help ensure that ALL children receive the timely diagnosis and care they deserve.

- **Find your senators' contact info here**
- **Find your house reps' contact info: here**

2. **Send an email** to your representatives through **this form**. All you'll need to do is input your information, and the email will be sent on your behalf!

Together, we can protect the future of newborn screening and ensure that every baby gets the best possible start in life.

ATTENTION CALIFORNIA PATIENTS

Do you struggle to access Hydroxocobalamin?

HCU NETWORK AMERICA

Organic Acidemia Association

Add your voice to a letter of appeal to The Medicine Shoppe regarding supply issues

bit.ly/4jcYUgJ

CHEMISTRY RX

ChemistryRx is enrolled with Medi-Cal and will ship to California!

bit.ly/3Y1U9y4

THE EVERYLIFE

RARE REPORT

News and Information to Educate and Activate the Rare Disease Advocate

#IAmMedicaid

Medicaid Cuts Threaten Rare Disease Care

Proposed cuts to Medicaid could put life-saving care out of reach for thousands in the rare disease community.

Many rely on Medicaid for treatments, diagnostics, medical equipment, home healthcare, and caregiving support. Cuts through the budget reconciliation process could limit access to these critical services as well as basic healthcare, especially for those depending on waiver programs to qualify for Medicaid.

There's still time to act. Tell your Medicaid story and share it with your Members of Congress. Your voice can make a difference, click below to take action.

TAKE ACTION

UPCOMING EVENTS

¡Una reunión virtual para pacientes y cuidadores hispanohablantes!

¡Regístrate aquí!

UPCOMING EVENTS

20% OFF

Select Promin Products

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Promo Code: PKUAWARE

Valid Through May 31, 2025

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In recognition of World HCU Awareness Day,
enjoy 20% off your order of select Promin products
purchased through the ZOIA Marketplace.

Stock up on nutritious and tasty low protein foods the whole family can enjoy!

zoiahealthcare.org

UPCOMING EVENTS

Save The Dates

Summer Regional MEETUP

Florida
June 7th 10:00 am-4:00 pm
Barnett Park 4801 W Colonial Dr. Orlando, FL 32808

Texas
June 28th 09:00 am-2:00 pm
May Valley Park The Woodlands, TX 77354

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IVDS
In Vitro Diagnostic Solutions

KNOW YOUR HCY

SIMPLY INSTANTLY ACCURATELY

IVDS is a clinical stage medical technology company with ambitions to develop the first and only meter to measure homocysteine in this way

IVDS INSIGHT

3 SIMPLE STEPS

SAMPLE COLLECTION
Insert a test strip into the meter and apply a tiny amount of patient's blood onto the test strip

ANALYSIS
Press a button and IVDS's proprietary technology gets to work, calculating HCY

RESULTS
HCY levels are sent directly to your phone within five minutes allowing you to make informed decisions in real-time

WE CARE ABOUT YOUR HEALTH

- IVDS was founded in 2017, to find innovative ways to diagnose and monitor metabolic disorders like HCU
- The HCY Now is currently undergoing pre-clinical testing to prove non-inferiority with mass spectrometry, considered the gold standard today

856-343-5098

ROBERT@IVD.SOLUTIONS

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 Michael to the table!

Michael joins us from Australia as we celebrate World Homocystinuria Day this month. He will share with us his families heartbreaking story to a diagnosis in honor of his late twin brother Phillip. As an adult patient with HCU, he has witnessed many advances and changes in diagnosis and treatment for this disorder and we look forward to hearing what his hopes are for the future of HCU.

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

Save The Date

July 10-12, 2026

HCU Network America
2026 Conference

Philedelphia, PA

It's A
FAMILY AFFAIR
In The City of Brotherly Love

HCU NETWORK AMERICA

Nexus
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Commited to making life a little easier for people with metabolic disorders through innovation, quality and customer service.

HCU EASY TABLETS

Easy To Measure ✓

11 HCU Easy Tablets equals 10g Protein Equivalent

Easy To Swallow ✓

Almond shaped tablets help for easier swallowing

Easy On The Go ✓

Convenient for formula consumption while traveling, at work, or at school

LEARN MORE

HCU Easy Tablets should be taken under strict medical supervision and are suitable from 3 years of age.
Nexus Patient Services now offers samples and patient assistance. Ask your dietitian for more information and a free sample.

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NPS-00032 April 2025

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

POWERED BY RARE X

The Homocystinurias affect multiple systems of the body!

The most common areas of the body affected are:

- The Central Nervous System (brain)
- Ocular (eyes)
- The Cardiovascular System (heart)
- The Skeletal System (bones)

But, many patients experience symptoms outside of these areas!

Help us to understand more:

Take the Health & Development Survey:
https://rare-x.org/homocystinuria/

CANPCU+ CANPKU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

Registration Open for flok Family Camps & Adult Retreat!

Registration is now open for our Adult Retreat and 2 terrific family camps for those with inherited disorders of protein metabolism, their family members, and friends. Learn more and register at flok.org/camp

- **Adult retreat**: A dedicated retreat for adults (21+) with IMD. $20 registration fee; travel support available.
- **Family Camp West** in Antelope, Oregon, June 5-8, 2025
- **Family Camp East** on Lake Winnipesaukee in NH, September 18-21, 2025

Thanks to support from BioMarin Pharmaceutical Inc. flok's retreat and camp are **FREE** for those with Classical HCU, MSUD, an Organic Acidemia, PKU, Tyrosinemia or a Urea Cycle Disorder. Flexible pricing available for family members and friends.

Questions? Contact camp@flok.org.

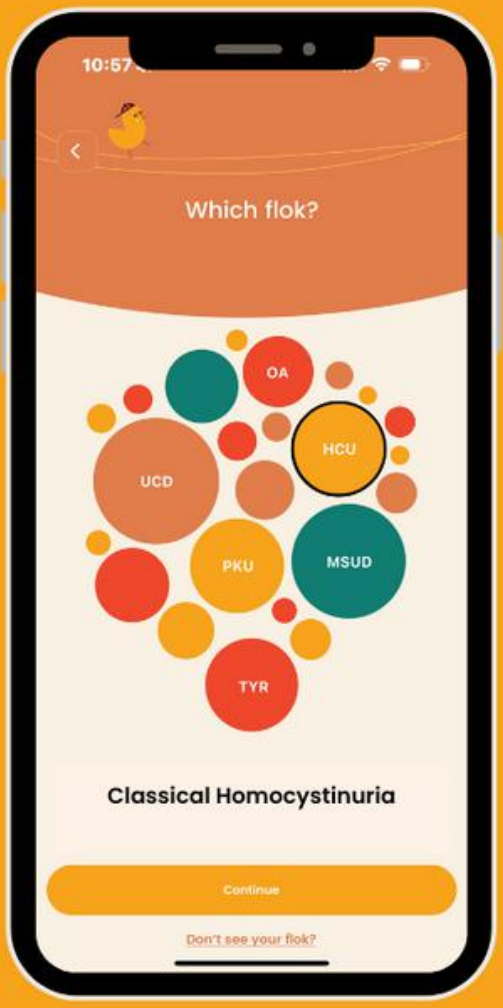
Start using the flok app today!

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.

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

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