The HCU Herald

Featuring...

HCU Hero

Nico from Scotland

June 2025

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

HCU HERO: NICO FROM SCOTLAND

Nico is our first of two children and was born in June 2019 in Aberdeen; which is located in the Northeast of Scotland. The pregnancy and delivery were a breeze (although admittedly this is being written by his dad...) and we were discharged the same day. As all new parents we spent the next few weeks navigating our way, mostly by trial and error, through the psychological enema that is the first exposure to parenthood. Despite our initial lack of parenting aptitude, Nico seemed very healthy, was easily breastfed and showed no signs of having any issues.

I am of Dutch descent, my wife is Scottish and neither of us have any known family history of inherited genetic disorders, metabolic or otherwise. Although Nico was subject to the routine newborn heel prick, the concept that Nico could be diagnosed with anything of that nature never even crossed our minds. Our lives changed, however, when he was around two weeks old, and we received a call out of the blue from the lead consultant in paediatric inherited metabolic medicine working for the Scottish National Health Service (NHS). It was a very short, and rather traumatic call, informing us that analysis of samples of Nico's blood had identified elevated levels of Methionine, was likely suffering from a rare condition known as Homocystinuria and that his untreated life expectancy would be around 35 years.

HCU HERO: NICO FROM SCOTLAND

Unsurprisingly, we were completely naïve as to what HCU was and after the short phone call, spent an emotional few hours frantically researching the condition online which only exacerbated our sense of panic. We were invited for a visit to the local hospital the same day where we met with paediatric clinicians who were able to explain in more detail, and with greater clarity, what the situation was and what our immediate options were.

“We were completely naïve as to what HCU was and after the short phone call, spent an emotional few hours frantically researching the condition online which only exacerbated our sense of panic.”

Further blood samples were to be taken to confirm the analysis, and Nico was to be subject to a two-week course of folic acid and pyridoxine supplements to test whether his measured amino acid levels would drop accordingly.

Nico showed no biochemical response to pyridoxine (B6), and he was officially diagnosed with classical Homocystinuria. We were asked to stop the treatment once this was confirmed and effectively switch his diet overnight from breastmilk to a specially formulated methionine-free formula (HCU Anamix Infant) as his measured levels were

remaining very high. Clearly it was not straightforward making the switch given the rather bitter taste of the formula but thankfully his blood levels (which were being measured on a weekly basis) were showing a rapid response to the treatment.

Once the dust had settled following a manic month or so including more blood tests, the failed treatment phase, switch to medicated formula and multiple road-trips to the metabolic centre in Glasgow we gradually began to realise what his only long-term treatment option was: a low protein diet accompanied by specially formulated powdered milk / paste protein supplement that he would take prior to every mealtime. We discovered early on that Nico is incredibly sensitive to natural protein and so his natural protein intake was, and still is, limited to 4 grams per day. In essence this is almost a negligible amount of protein, and the result is that he cannot really eat anything natural above and beyond fruit, select vegetables and small snacks. This is unfortunate but from purely a treatment perspective it does simplify matters, particularly when instructing others as to his dietary requirements.

Both my wife and I work full time and so since the age of one Nico has been at childcare (and now school) five days a week from 8am-5pm. Given the rarity of the condition my wife has spent many hours, with the help of literature provided by the health service, educating the childcare staff on his diet and treatment (emphasising that it is not an allergy as that would result in meal-time isolation from his peers).

HCU HERO: NICO FROM SCOTLAND

In addition to the low protein processed food that provides the bulk of his calories, Nico has always been happy to snack on low protein natural foods like fruit and certain vegetables.

We are very fortunate that Nico's low protein food comes prescribed through the Scottish NHS, with items such as low protein pasta's, sausage / burger equivalents, bread, egg replacement etc. sourced from various countries in Europe as well as the United Kingdom. His medicated protein paste is made by Nutricia (owned by Danone) in the Netherlands. Given my heritage we regularly travel to the Netherlands and one day I intend on asking if we could have a tour of the factory that is so important to us.

Throughout his life Nico has mostly been pretty good at eating his low protein food and protein supplement and from a very early age appeared to understand that he must be selective with his choice of food: often rejecting anything offered to him by non-family members whilst referring to his 'special belly'. The only serious wobble we have had to date was when he was two (of course....) and he refused to touch his protein paste for three weeks. Although looking back it seems rather innocuous and just normal boundary testing behaviour of a typical two-year-old terror, at the time we were riddled with anxiety and dreaded the worst. Worried that he would become protein deficient, I ended up building a spreadsheet detailing methionine percentage as a function of total protein content for hundreds of food types to understand what natural protein we could feed him whilst keeping his methionine levels supressed.

HCU HERO: NICO FROM SCOTLAND

Thankfully it never came to that as he figured three weeks was sufficient torture time for his parents and gradually began to eat his paste again.

These days Nico is a very sociable, active child and understands the key aspects of his condition. He enjoys going to school and participates in activities like swimming, football (soccer), rugby and tennis. We take monthly blood samples at home (which usually requires a few bribes) and deliver them to the hospital for analysis. His height and weight are monitored, and his eyes are checked on an annual basis for any abnormalities. His blood levels have remained relatively stable since we started his treatment, and he has not displayed any of the symptoms associated with untreated HCU.

Nico has an uncomplicated approach to life with his only objective for each day to have as much fun as can be squeezed into 14 hours (often waking his sister and parents up in the early hours in order to get a head start on that daily goal....). When we travel, we bring a suitcase of food for him and when we go out to restaurants and / or parties we try and make something in advance that will be similar to whatever type of food others will be consuming. We have reached an agreement with his school that they will prepare three meals a week for him (for which we are very grateful) on the basis that we provide the raw ingredients. To date we have had no issues with any of these, clearly the increase in niche diets globally has raised individual awareness and understanding of the need for bespoke dietary requirements.

HCU HERO: NICO FROM SCOTLAND

Our main challenges regarding his diet for the coming years will be ensuring we gradually introduce more varied and complex tasting food into his routine as his pallet evolves and hopefully allow him to get some satisfaction out of eating rather than seeing it as a necessary chore.

Additionally, as he gets older, he will gain more responsibility for his own consumption, and it is probably only natural for us to worry about what decisions he chooses to make when he does get to that stage (as every parent probably does!). Interacting with other, similar aged, HCU patients as a teenager and beyond will hopefully help guide him as he makes the journey into adulthood.

Nico has a younger sister, Naomi, who was born in 2021 and does not suffer from Homocystinuria. She was subject to more focussed testing upon birth due to the high likelihood (25% chance) of having the condition and we were clearly delighted when the blood results returned negative. The moment was a little bittersweet though as the natural elation we felt knowing Naomi was in

the clear was a subconscious acknowledgement, combined with some repressed guilt, of the impact the condition will have on Nico's life.

HCU HERO: NICO FROM SCOTLAND

The condition is very rare in Scotland as Nico is currently (we believe) only the third child to have had the condition diagnosed since it was added to the heel-prick screening list (we were very fortunate as HCU was only included a few years prior to his birth). When Nico was born, he was the second child in Scotland diagnosed with the condition and we were very grateful that the parents of the first child were willing to

talk to us and discuss their experience and likewise we spoke with the parents of number three. The community nurses, metabolic specialists and paediatric dieticians working for the NHS have been invaluable to us over the years, providing guidance and advice, ensuring all his supplies are delivered in a timely manner, monitoring his development and helping with numerous other tasks that have allowed us to maintain control of his condition. We have no contact with any other HCU patients in our area and joining networks such as HCU America will hopefully allow us (and eventually him) to engage with a large community of people in similar situations. Unfortunately, DNA testing was not able to identify all the mutations responsible for his HCU, but we still hope that alternative treatment methods become available in the future and, if so, we could potentially carry out further DNA tests to better understand (and possibly treat) his specific condition. We will keep a close eye on ongoing studies and research.

Share Your Story

With Us!

To Join or Learn More:
Email
info@hcunetworkamerica.org

JOIN OUR FUNDRAISING TEAM

Help create, organize, and support new and existing fundraising ideas!

These virtual funds help support our outreach, programs, and research!

To Join or Learn More:
Email
Dbartke@hcunetworkamerica.org

WHAD RECAP

World Homocystinurias Awareness Day *is an initiative of patient organizations and consortia globally who have a focus on the homocystinurias. The main objective of World Homocystinurias Awareness Day is to raise global awareness of the homocystinurias and their impact on patients' lives.*

Hope Connects Us

World Homocystinurias Awareness Day 2025

Around The World

Thank you

to all who participated by spreading awareness and by participating in our Raffle Fundraiser or making a Donation!
We were able to raise $6,240 for our Education & Outreach programs!

Syntis Bio

HCU NETWORK AMERICA

RAFFLE WINNERS

WINNER: JUNE ANDERSON

WINNER: LIZ CARTER

WINNER: LOUIS GASPERINI

WINNER: HOLLY FULGINITI

WINNER: JUNE ANDERSON

WINNER: LINDA CONARD

JUNE EVENTS

Scan to Register

Scan to Register

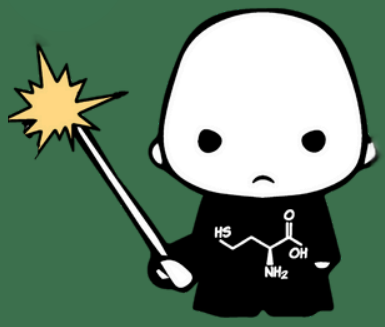
THIS SEPTEMBER

HCU HEROES

Race for Research

CURE

WALK • RUN • RIDE



Cystadane; the last FDA approved drug for the homocystinurias (HCU) was approved in 1996. That was 29 years ago!

Currently only two clinical trials are in progress for classical HCU, but there are none in progress for cobalamin disorders and Severe MTHFR.

In collaboration with other organizations, HCU Network America has issued 10 research grants.

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

https://bit.ly/HCURace25

Pricing

- The first 100 registered receive a medal
- All participants will receive a shirt (Deadline August 18)
- Early bird pricing - $20
- After July 15 - $30

RECORDATI RARE DISEASES
Focused on the Few®

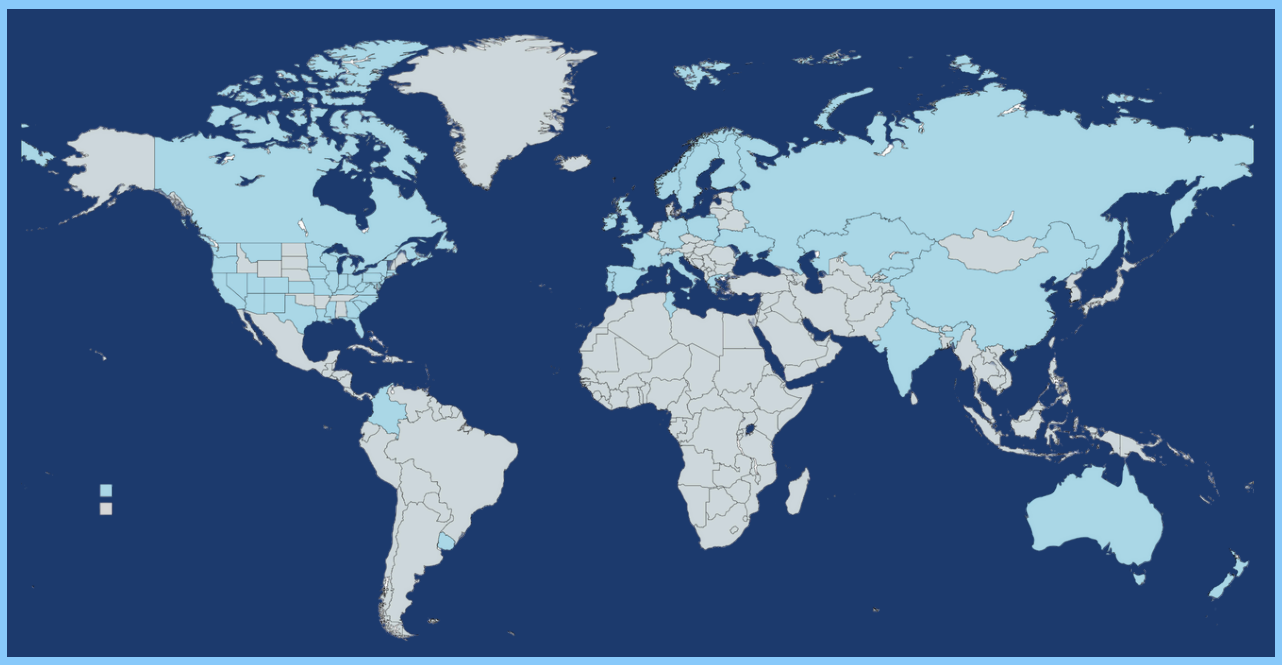
HCU Network America is a registered 501c(3) foundation, registration number 81-3646006. All donations are tax deductable.

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

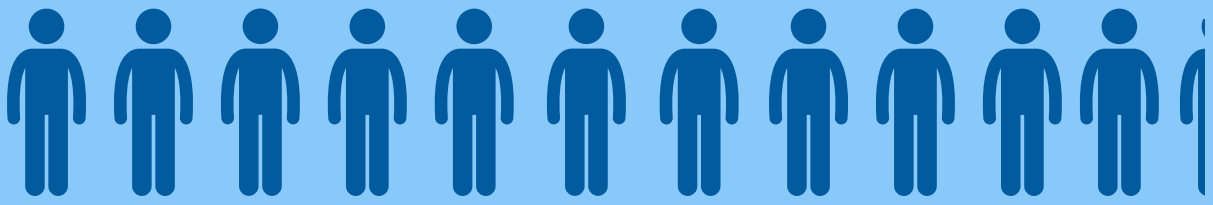
POWERED BY RARE-X

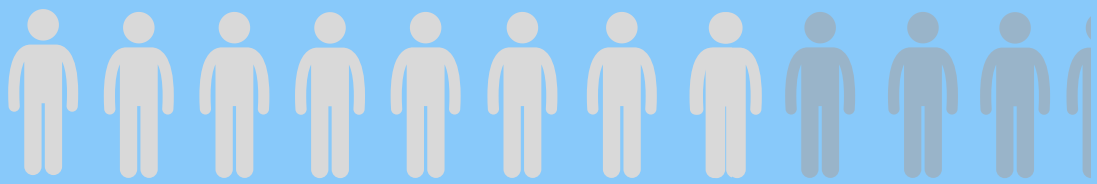
114 PATIENTS

23 COUNTRIES AND 35 US STATES



114 people have enrolled in the HCU Data Collection Program (DCP)





Only 76 have completed the Head to Toe Survey **Without you completing this survey, your data is incomplete!**

Click here to enroll/complete your surveys

CANPCU+ CANPKU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

Incase you missed it...

The HC&U Podcast has new episodes every month!!!

Check out the WHAD Bonus Episode!

Meet your hosts!

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

To Listen:

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

LISTEN ON Spotify

Listen on Apple Podcasts

LISTEN ON iHeartRADIO

Listen on amazon music

The latest episode

In this episode, Ben welcomes 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.

Save The Date

July 10-12, 2026

HCU Network America
2026 Conference

Philadelphia, PA

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

HCU NETWORK AMERICA

Summer Recipe: Veggie Burgers

Ingredients:

- 1 large zucchini, chopped
- 1 medium carrot, peeled and chopped
- 1 small onion, peeled and chopped
- 1 cup sweet potato, peeled and diced
- 1 tbsp fresh parsley, chopped
- 2 tsp dried mixed herbs
- Salt to taste
- Black pepper to taste
- 1 ¾ cups (200g) low-protein baking mix, divided
- 2 tbsp olive oil, divided

Instructions:

- Place vegetables into a food processor and pulse until finely grated and combined.
- Place vegetables into a mixing bowl with herbs, salt and pepper. Stir to combine.
- Add 1/2 of the low protein baking mix to the vegetables, stir to combine. Add remaining low protein baking mix; gently stirring until it is well combined with the vegetables.
- Dust hands with a little of the low protein baking mix. Gently take the vegetable mixture out of the bowl and shape into a large ball. Cut in half, and then quarter each half. You will now have 8 even-sized portions.
- Heat 1 Tbsp of oil in a medium-sized non-stick frying pan. Take one of the vegetable portions; roll it into a ball, slightly flatten, and place into the pan. Repeat with 3 more portions. Cook for 2–3 minutes; until golden brown. Flip each, cooking the other side for 2–3 minutes. Remove vegetable patties from pan and repeat with the remaining uncooked vegetable portions.

Nutrition

Serving: 1 Patty

Calories: 160

Protein: 1.3 g

Recipe Makes: 8 servings

Prep Time: 15 minutes

Cook Time: 10 minutes

Vitaflo™

UPCOMING EVENTS

pku ORGANIZATION OF ILLINOIS

SPONSORED BY NPKUA national PKU alliance

ILLINOIS LOW PROTEIN FAMILY CAMP

July 10 – 13th, 2025

Join us back at the beautiful Great Oak Retreat Center for a weekend away with other PKU and IEM families. Memories will be made, meals will be shared and friendships will be fostered.

FUN ACTIVITIES & GAMES FOR THE WHOLE FAMILY!

- SWIMMING + FISHING
- CANOEING + PADDLE BOATING
- ARCHERY + ZIPLINING
- AND MORE!

GREAT OAKS RETREAT CENTER
1380 COUNTY RD 900 N
LACON, IL 61540

$40 / person
$100 / family

SCAN ME TO REGISTER!

Information : dbartke@hcunetworkamerica.org
pjkowalczyk@yahoo.com

Click Here to Register

BUILDING BRIDGES

Family Conference

August 22-24, 2025 - Calgary, Alberta

Whether you're newly diagnosed or have been navigating this journey for years, this event offers connection, education, and encouragement in a welcoming and supportive environment.

Throughout the weekend, families will have access to:

- **Educational Sessions & Breakouts** led by clinicians, dietitians, researchers, and community leaders on topics like treatment updates, daily management, mental wellness, advocacy and more.
- **Low-Protein Meals & Snacks** provided throughout the weekend, ensuring safe and inclusive food options for attendees with dietary restrictions.
- **Onsite Childcare** for children 12 and under, giving parents the opportunity to fully participate in sessions while their kids engage in fun, supervised activities.
- **Teen Programming** tailored for youth, with peer connection, age-appropriate sessions, and interactive learning.
- **Vendor Tables** showcasing low-protein products, supportive services, advocacy groups, and community resources.
- **Social Activities** including an optional Friday Dinner Gala, Saturday's family-friendly pool takeover and informal meetups to connect with other families across Canada (and North America!).

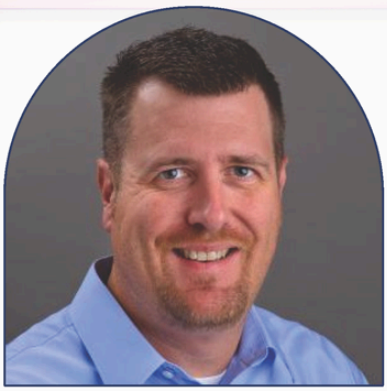
www.canpku.org/BuildingBridges for more information

BUILDING BRIDGES

Speakers

Dr. Kim McBride, MD, MSc, FRCPC, FACMG

Dr. Beth Potter, PhD, MSc,



Dr. Shawn Christ, PhD

Other Experts include:

- **Dr. Fernanda Leal-Pardinas, MD, MSc**
- **Dr. Nerissa Kreher, MD, MBA**
- **Dr. Gail Ouellette, PhD, MSc, CGC**
- **Alexander Lim, MPH, LLM**
- **Susan Marlin, MSc**
- **Dr. Beth Potter, PhD, MSc**
- **Dr. Iveta Sosova, PhD, DABMGG, FACMG**
- **Sophia Khan,MSc, PhD student***
- **Madison Fennell, MSc student***
- **Catherine Rombough, RD along with other Registered Dietitians**
- **Lynn and David Paolella (Cambrooke)**
- **HCU Network America**

.... and still more to come!

Contact Register

What is the contact register?

The contact register is a secured private survey that allows you to share information on you or your family member with HCU with us. This includes where you are from, your relationship to homocystinuria, the patient's birthdate, gender, their exact diagnosis (e.g. CBS, cobalamin, or MTHFR), how they were diagnosed, and if the patient was diagnosed through newborn screening. This information is kept confidential and will not be shared unless you give us permission. By registering, you will be able to identify other patients in your state and request their contact information. You will also be able to access information posted over time that can only be shared with the patient community. (For example, we may have webinars that the expert presenter does not want to be publicly available, but is willing to share with the HCU community.)

What will this information be used for?

HCU Network America strives to inform patients and families with resources, create connections, and support advancement of diagnosis and treatment of HCU and related disorders. The information you provide helps us succeed in our mission – plan events, develop resources and educational tools, and ensure everything is being done to support timely and accurate diagnosis from birth. It also allows us to have informed conversations with doctors, pharmaceutical companies, and law makers. Your information helps us understand the landscape better so we can better advocate for you!

How do I participate?

The contact register form takes approximately 3-5 minutes to complete. You can find the form either by visiting our website and clicking on the "Contact Register" tab, or you can fill it out by going directly to: **https://www.surveymonkey.com/r/HCUContact**

FOLLOW US

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