The HCU Herald

Featuring...

HCU Hero

Olive From the United Kingdom

July 2025

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

HCU HERO:
OLIVE FROM THE UNITED KINGDOM

Matt and I welcomed our first child, Olive, into the world on 28th March 2023 after a long labour and uneventful pregnancy. She was, and remains, the most perfect and beautiful little girl we have ever seen. Everything seemed relatively straightforward in the first few months, and we were fully immersed in the newborn haze and overwhelming love for our daughter. It is with hindsight now there are some things in those early stages which may have been the first signs that something wasn't quite right.

Shortly after birth, Olive started vomiting very bright yellow liquid, but doctors didn't seem concerned, and it soon settled. Despite seeming to breastfeed fairly well, she also wasn't putting on weight as quickly as you'd expect. From about 8 weeks her poo totally changed colour and consistency, becoming various shades of green and runny. I took her to see the GP on numerous occasions with concerns about various things but was always told that she was a healthy baby.

As a first-time parent, you always want to believe this is the case. I even found myself at a friend's wedding thrusting photos of Olive's poo in front of someone I discovered was a pediatrician to see if he had any opinions on what could be causing it!

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Developmentally, there didn't seem to be anything out of the ordinary; Olive rolled and smiled in line with what is considered "normal", so we continued enjoying our life as a new family.

It was around 6 months that Olive started to vomit daily. The volume was alarming, and it varied from being milky to totally clear liquid. Back to the GP we went, but without witnessing one of the vomiting episodes, my concerns were again dismissed as being normal - all babies are sick after all. We knew her vomiting was far from normal and ended up seeking help from a private gastroenterologist, who suggested it could be an allergy or reflux. When an elimination diet and medication didn't put an end to it, we felt lost as to what it could be and what to do next.

We also started to have concerns around Olive's development. It became apparent that Olive was somehow different from babies in her peer group, and she wasn't meeting expected milestones. Consequently, I started to withdraw from social situations where I would see other babies, I found the constant comparing and talking about what my baby was doing demoralising.

Olive also didn't strike me as a particularly happy baby. I can recall very few times within her first year when she laughed, and I didn't feel she was especially attached to me, despite us spending all our time together and obsessively breastfeeding her as my concerns around her weight gain continued.

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She was increasingly unsettled, and during a trip to Australia over Christmas at 9 months old, I became more concerned about her size. Every time I held her, I was alarmed at how light she was. She was also becoming increasingly lethargic - we needn't have dreaded the day-long flight over to Australia, she slept for most of it.

On our return to England in January 2024, the realisation that something was gravely wrong with Olive was starting to dawn.

Our attempts at weaning her were unsuccessful, and I felt the pressure on breastfeeding her overwhelming at times as she continued to refuse a bottle. At a pediatric appointment at our local hospital, we flagged concerns regarding her meeting milestones, lethargy, and aversion to solid foods. By this stage, I was also fearful about having her weighed and wasn't able to be in the room whilst it was done. My fears were recognised when this latest result was plotted on the centile chart, and she had fallen off the chart entirely.

She was recommended for a blood test (and this would be her first), but I was alarmed that he didn't seem otherwise concerned about her. Again, whilst trying to take this as a positive, there was a sinking feeling that something was very wrong.

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I will confess I was always Googling Olive's symptoms. I often felt in the absence of medical help, Google would give me the answers. It was between this last appointment and her blood test that I came across the symptoms of B12 deficiency and Olive presented with all of them.

When we went for her blood test, I begged her phlebotomist to test her for it, but it hadn't been specified by the paediatrician. I ended up in tears with the receptionists asking for it to be tested, but I was met with the words "if her doctor felt it was necessary, it would be tested". Hours later, I got a call from her doctor saying we had to come into the hospital immediately as her blood test results had come back and there was reason to believe she could have leukaemia because of her white blood cell and platelet counts.

I sat, frozen in a café with Olive in my arms; she looked the palest and fragile she had ever looked, and I felt as if she'd just been handed a death sentence. We travelled to the hospital in total disbelief, and I had to recount her entire medical history before repeat blood tests were done. After an agonising wait, the initial results, which had caused the most concern, had normalized, but they identified that she had macrocytic anaemia and elevated ALT.

We stayed in overnight, feeling slightly reassured, until the next morning when another blood test was performed to ensure that the normalised result wasn't the anomaly, and, thankfully, it wasn't. It was whilst here that I asked if B12 had been tested, and they said it was within normal range but on the lowest end. Her strange results (persistent macrocytic anaemia and a fluctuating thrombocytopenia) prompted them to be sent off to another hospital, which specialised in paediatric haematology.

HCU HERO: OLIVE FROM THE UNITED KINGDOM

In the meantime, she was fitted with a feeding tube and put under the care of a dietitian to monitor her weight, and we were trained in giving her top-up feeds of formula to supplement the breastfeeding. We found the feeding tube quite daunting initially, but it was a huge relief to be able to give her extra calories and nutrients.

We met with the haematologist and, although she didn't really have any answers, she did say that she had macrocytic anaemia and recommended a course of fortnightly B12 shots to get her levels up. The possibility of it being a metabolic condition was mentioned around this time, but it was just considered one of many possibilities. Olive was given her first dose of B12, and we would come back after a fortnight for the next.

There was an immediate improvement in her energy levels, her vomiting stopped, and she was even making efforts to crawl. We weren't sure whether it was down to the B12 or the extra food she was getting, but it was truly amazing to see such an instant improvement, and at a repeat blood test, there was haematological improvement as well. Olive was also referred for an abdominal ultrasound to check her liver to see if a cause for the raised ALT level could be identified, but she was given the all-clear.

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Given ongoing issues with feeding, we decided to also seek the help of a private speech and language therapist, and one thing I was interested to find out was whether she might have an undiagnosed tongue tie, which could be causing all her feeding issues. And apparently, she did!

This felt like a strange sort of relief – perhaps all her problems were down to this. Oddly, the specialist called me out of the blue one evening to say that she thought Olive's eyesight was a bit "off" and asked if we had it checked out. I noticed her lack of ability to focus, and it was also around this time that she started to lose the ability to sit independently or bear weight on her legs.

Following the tongue tie diagnosis, we were eager to get it cut and I couldn't wait to see how things would change after this appointment. Sadly, it was the night of her tongue tie cut that Olive had her first eye-roll episode and all the hopes for a fresh start were quickly dashed. Initially, we thought it might be related to the stress of the day but over the next few days she continued to have repeated episodes. Each lasted a few minutes, and she seemed fully conscious throughout, but it felt totally abnormal.

When I spoke to her paediatrician about the eye rolling and regression in skills, he put it down to her anaemia. To my shock, she said that parents too often brought their children into hospital with eye rolling and it was just tiredness. Again, I tried to take this conversation as a positive, but I felt far from reassured.

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Olive continued with eye rolling episodes, and after a week we took her to A&E. We were on the brink of being discharged home, having been reviewed, when we assume something in her medical history stood out to one of the doctors and we were kept in overnight with an MRI was scheduled the next day. They wanted to attempt this without sedation, and so we were locked in a tiny, dark room whilst we tried to get her to sleep. Inevitably, the whirring of the machine woke her up, and the MRI was unsuccessful, so we were sent home with an EEG booked for the next day. It was during this that I recall another heart-stopping moment as a consultant was called in to review the screen as she was monitored, a far from promising sign that things were ok.

We were sent home and told someone would be in touch, but before we even got that phone call, we were back at the hospital as an eye-rolling episode was then accompanied by Olive collapsing her head to the side repeatedly, which was truly terrifying. We were told that the EEG had demonstrated hypsarrhythmia, and Olive was epileptic.

We would be transferred to a hospital with a specialist paediatric neurology department so they could explore this further; the following days would be some of our darkest. Recounting Olive's complicated medical history and reeling off her myriad symptoms to the neurologist was further proof that there must be something underlying that was causing so many problems. They repeated the EEG, which showed the same result.

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They were confident she was experiencing Infantile Spasms. She was booked in for an MRI under sedation, and kissing goodbye to Olive in her tiny, fragile state as she was put to sleep was utterly heart-wrenching. After an agonising day of waiting for the results, they thankfully came back normal, apart from delayed myelination, which they said could be rectified in time.

Olive was started on an aggressive course of steroids and anti-seizure medication to try and stop the epilepsy before it could cause any longer-term damage. The seizures did stop, but the drugs totally changed her and took away any traces of her personality. They did, however, make her ravenously hungry, and we relished seeing her eat food enthusiastically for the first time! Before being discharged, we were given the option to take part in a study called Gene-STEPS, a global research study through Great Ormond Street Hospital that used rapid genome sequencing to provide early detection of the cause of epilepsy in babies under one. Blood would be taken from Matt, Olive, and me to look for changes in the child's DNA that might explain their condition. Without hesitation, we said yes and told the results could be back as quickly as a few weeks.

We returned home and adjusted to the new medication routine, a dramatically changed almost-one-year-old, and uncertainties around our future. After two weeks, her neurologist called with the results, saying they had found a defect with her MTR gene leading to a diagnosis of Cobalamin Disorder G, and we would be contacted by Great Ormond Street Hospital to come in to start her B12 treatment immediately. My overriding emotion on receiving this news was relief – there was a diagnosis, a treatment plan, and finally some answers. The call from the hospital came the next day, and off we went.

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A team from the metabolic department sat with us for over an hour, explaining her diagnosis of cblG, its rarity, and that Olive presented as a ‘textbook case’ for the disorder. They were reassuring, knowledgeable, and said that they hoped she was on the milder side given her presentation. She received her first injections (10mg/2ml of hydroxocobalamin), and we were trained in administering them ourselves.

Alongside folinic acid tablets, this would be the medication she would receive daily. Olive was discharged on her first birthday and after two days of treatment, we already started to see changes in her. She engaged in books and was happier and much more energetic. A brighter future finally felt within grasp.

Over the next few months, Olive weaned off her anti-seizure medication, and betaine was introduced. The strides in her development were startling. Whilst we have the medication to thank for this, we also attribute so much to Olive’s resilience and determination. She took her first steps at 20 months and words followed shortly after.

Olive recently celebrated her second birthday, and it allowed us to reflect on how far she (and we) had come in a year, and the challenges we’d faced as a new family. It’s impossible to articulate how proud we are of her and all that she has achieved, despite the setbacks.

Her doctors continue to be astounded by her progress and, though we have no doubt there will be bumps in the road, we are hopeful that she will live and full and happy life and not be defined by her condition.

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Her homocysteine levels are consistently around the 20 mark and all other levels are normal and, apart from some mild speech delay, we are no longer concerned about her development.

To any parent facing a new diagnosis or who has experienced a similar journey to us, keep going - you know your child better than anyone else, and there is a lot to be said for pure parental instincts.
Today, Olive is an energetic toddler who prefers to run rather than walk, picks up new words every day, can sing along to her favourite songs, and loves nothing more than a cuddle! We are so glad we found HCU Network America and connected with families who can guide, advise, and above all, reassure. I hope Olive's story might do that for another family someday.

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

UPCOMING EVENTS

Adulting 101: Find Your Path
With HCU & MSUD

Presentation & Panel | Sept. 11 at 6 pm ET

Jessica Gold, MD, PhD
Northwell Health

Topics Covered:

- Self-Management
- Navigating School, Work, and Routine
- Healthcare and Appointments
- Social Life and Relationships
- Independent Living and Life Skills
- Education, Career, and Future Planning

As teens and young adults living with **classical Homocystinuria (HCU)** or **Maple Syrup Urine Disease (MSUD)**, the path to independence comes with unique challenges—and triumphs. This collaborative webinar from **HCU Network America** and the **MSUD Family Support Group** shares real stories and practical insights on growing up, taking charge of your own care, and thriving with a rare metabolic disorder.

Register here: https://bit.ly/Adulting101-25

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UPCOMING EVENTS

EVERYLIFE FOUNDATION FOR RARE DISEASES

15TH ANNIVERSARY

RARE ACROSS AMERICA

VERY VOICE, IN EVERY DISTRICT, MATTERS

Aug 4-15 2025

Donate

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

Where?

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

When?

Meetings with Members of Congress will take place between August 4th and August 15th. We will be offering training webinars to help advocates prepare for their meetings.

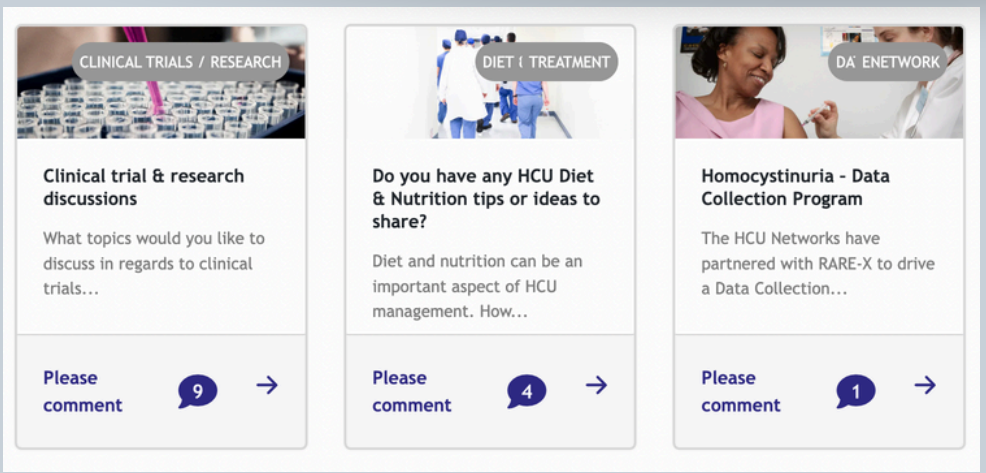
- July 15, 2:00-3:00pm ET: General Training Webinar
- July 22, 3:00-4:00pm ET: Team Coordinator Webinar
- July 23, 4:00-5:00pm ET: Share Your Story With Policymakers Webinar

REGISTER HERE

JOIN THE HCU E-NETWORK

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/

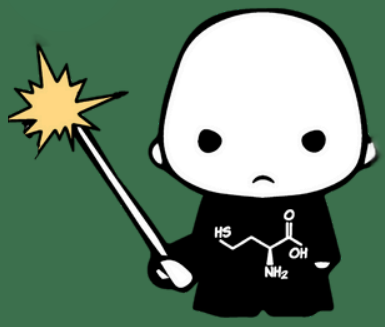
THIS SEPTEMBER

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

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Focused on the Few®

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

IN CASE 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

Ben welcomes Allison to the table!

Allison is a Senior Policy Analyst at the National Organization for Rare Diseases. Ben and Allison discuss the state of newborn screening in the United States. They also dive into the discontinuation of the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). Lastly, they will explore what the future looks like for newborn screening.

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

Information : dbartke@hcunetworkamerica.org

Click Here to Register

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

POWERED BY RAREX

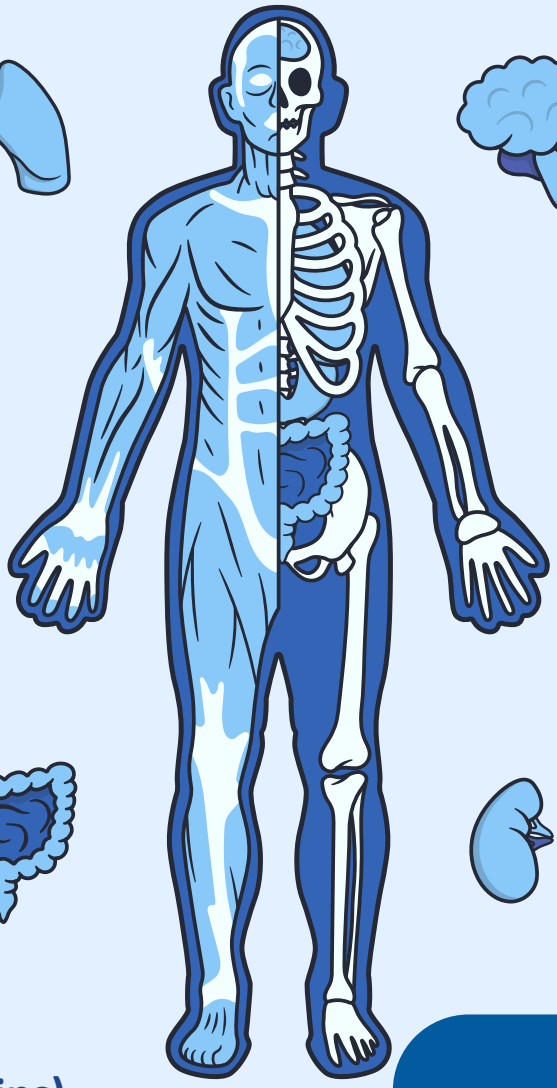
Head To Toe
WE WANT TO KNOW

Skeletal System (Bones)

Central Nervous System (Brain)

Ocular (Eyes)

Cardiovascular System (Heart)



Gastrointestinal System (GI Tract)

Digestive System (Stomach)

Vascular System (Veins)

Click Here
To Take The Head to Toe Survey

CANPKU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RAREX

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*!

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

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!).

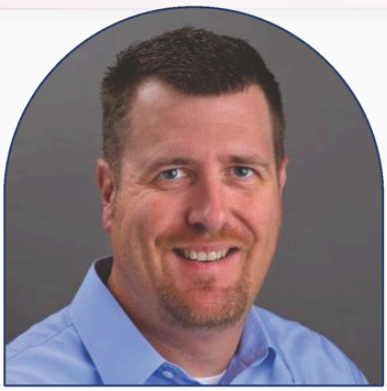
www.canpku.org/BuildingBridges for more information

BUILDING BRIDGES

Speakers

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

Dr. Beth Potter, PhD, MSc,

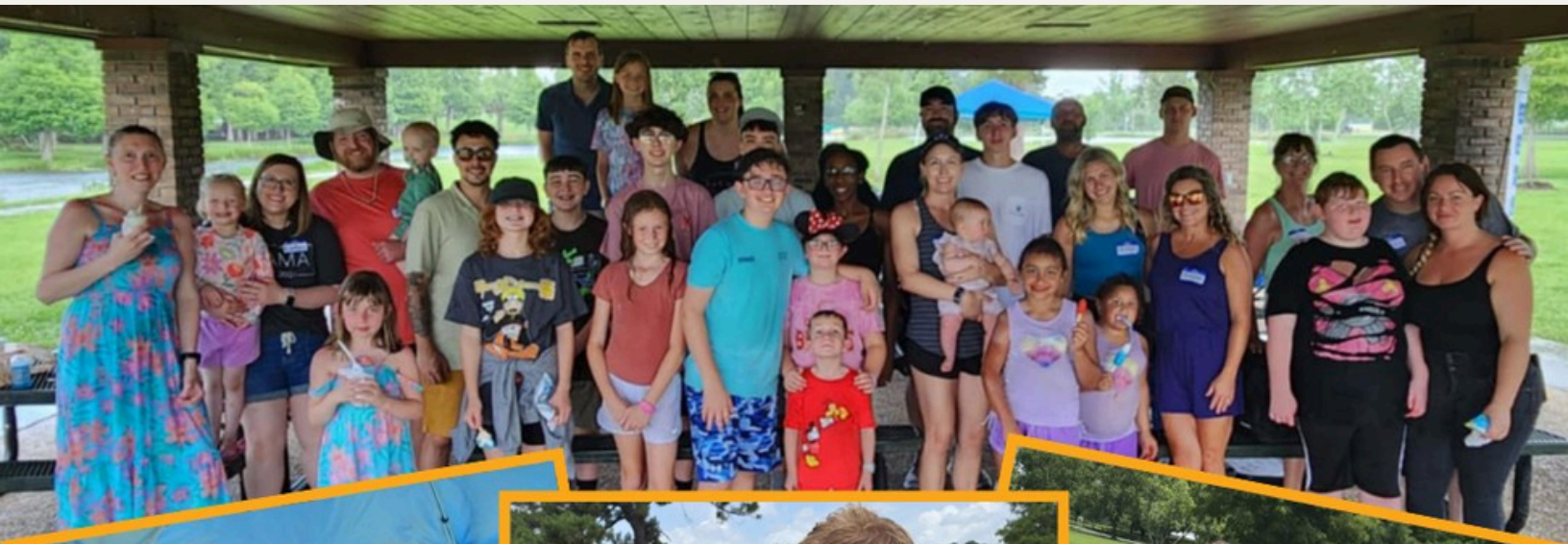


Dr. Shawn Christ, PhD

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!



Florida Summer MEETUP RECAP

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eTon PHARMACEUTICALS

NUTRICIA

TRAVERE THERAPEUTICS

Texas Summer MEETUP RECAP

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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

FOURTH OF JULY FLAG CAKE

Protein: .4 grams
Serving Size: 1/12 Cake

Ingredients

- **CAKE:**
  - 1/2 cup sugar
  - 1/2 cup butter, softened at room temperature
  - 1 ½ cups low protein allpurpose baking mix
  - 2 tsp baking powder
  - 2/3 cup almond milk, unsweetened
  - 2 tsp egg replacer
  - 2 tsp vanilla extract
  - 7 medium-sized strawberries, fresh, sliced
  - 25 small blueberries, fresh
- **BUTTERCREAM ICING:**
  - 3/4 cup butter, softened at room temperature
  - 2 cups confectioners (powdered) sugar
  - 1-2 Tbsp almond milk, unsweetened
  - 1/4 tsp vanilla extract

Instructions

CAKE:

- Preheat oven to 360° F.
- Cream sugar and butter in a medium-sized bowl.
- Add low protein baking mix and baking powder; rubbing together with your hands until it resembles breadcrumbs.
- In a separate bowl combine almond milk, egg replacer and vanilla.
- Add the wet mixture to the dry, and mix until well combined.
- Pour batter into a parchment paper-lined 7 inch square cake tin, and bake in the preheated oven for 15 minutes, or until toothpick inserted in the center comes out clean.
- Remove from oven, place on a cooling rack.

BUTTERCREAM ICING:

Beat the butter in a large bowl until soft. Add half of the powdered sugar and beat until smooth. Add remaining sugar, one tablespoon of almond milk, and vanilla, beating until creamy and smooth. Beat in the remaining milk, if necessary, to reach your desired consistency.

TO DECORATE:

- Spread 3/4 of the icing on top of the cooled cake.
- Make a square with the blueberries in the upper left corner of the cake.
- Place a row of strawberries across the top of the cake like a red stripe.
- Add the remaining icing into a pastry bag fitted with a star tip, and pipe a row of white stripes below the strawberries.
- Alternate rows of strawberries and icing until the flag is completed.

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