# The HCU *Herald*

*Featuring...*

## HCU Hero

Mason from Texas

June 2026

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

# HCU HERO: MASON FROM TEXAS

Mason has Cobalamin C and it was discovered through the newborn blood screen process.  His vision was perfectly normal when he was born, but macular degeneration started developing when he was one month old.  I can very vividly remember the day when we were told that vision loss had started and that Mason was going to be legally blind.  One of the first thoughts I had was that he's not going to be able to do any of the fun things that kids get to do growing up. Looking back at all the amazing things Mason has done, that was a horrible, negative thought and couldn't have been further from the truth.

My wife and I have always done our best to treat Mason the same way that we treat his older sister Madyson.  We encourage them both to be active and find hobbies that they love to do.  One of the benefits of raising our children in Wyoming is that the towns are close-knit communities; they support kids' sports, and are parent coached.  Soccer is one of the first sports that kids can try, so of course we signed Mason up.  We gave the city's recreation center a heads up that Mason had low vision, and they asked if we needed to get special soccer balls that blinked light or make noise. Based on Mason's vision and the fact that the soccer balls were bright yellow, we didn't feel it was necessary, but it made us aware that there are always options out there.

# HCU HERO:
# MASON FROM TEXAS

Mason had tons of fun playing soccer with his friends, and the coaches always made sure that he was included and got to play. They would even set things up so that he could score a goal.

Mason was also heavily involved in Cub Scouts in Wyoming.  Like soccer, several of Mason's friends and classmates joined Cub Scouts when he did.  Scouts in Wyoming was a great experience and taught the kids a lot of great skills that they will use throughout their lives.  Mason's den leader did a phenomenal job making Mason feel involved. She would enlarge materials for him and always made sure he was included in all the activities. Mason has earned his Arrow of Light, the highest award that a Cub Scout can earn, but he's chosen not to move on and join the Boy Scout troop.  Before he made that choice, we did some research.  We found out that scouts with special needs can work with their council and build a plan with accommodations so that they can become Eagle Scouts.  The process is very similar to the IEP process in school for setting accommodations

# HCU HERO:
# MASON FROM TEXAS

and making sure that everyone is aware of them and most importantly making sure they are successful.

Our family has always been huge fans of baseball.  In our small town of Wyoming there was an adaptive baseball program through Little League called the Challenger League.  It is an adaptive baseball league for children and young adults with special needs.

It's a great experience because each player can have helpers with them to help run the bases and play in the field.  The helpers are usually volunteers from the baseball teams, softball teams, scouts, local honor societies, and many other local groups. They don't keep score in the games, but they make sure that every player gets to bat and field the balls. The leader of the Challenger League found another special needs baseball program that had some other benefits including a field made with astro turf specifically designed for wheelchairs.  Each year the Miracle League puts on an All-Star weekend and 2 players from each team are allowed to go. In 2025 Mason was able to represent Wyoming and it was an amazing time.  The entire town of Covington, GA welcomed all the special needs families and made everyone feel so special.  It is so amazing to watch these kids have so much fun playing America's past time.

# HCU HERO:
# MASON FROM TEXAS

In the summer of 2025, Mason found his passion.  Mason's Orientation and Mobility teacher held a summer camp for all the low vision kids in the district.  They did amazing things like scavenger hunts at the local grocery store. They would find items on a grocery list and make sure they didn't overspend their budget.  It was great because it taught the low vision students critical life skills like scanning the aisles and shelves, something that we take for granted.

One of the best things that they did in this low vision summer camp was a 30-minute taekwondo class each morning. Kwan Jang Nim (Grandmaster of ICON Martial Arts Academy) was honored to be able to have a Taekwondo class with these special students. He knew they would potentially have issues with balance, core strength and the ability to watch the demonstrations of the taekwondo techniques, but he adapted his training style and educated his assistant leaders on the best ways to teach these students.

Taekwondo isn't just about punching and kicking, it's about being strong mentally and spiritually.  Taekwondo isn't about being the best in class, it's about being the best that you can be.  The students learned so much in those few short days, and it was truly inspiring to see what they were capable of. Kwan Jang Nim enjoyed his time so much with them that he started a class for these low vision students called the VIP class.

# HCU HERO:
# MASON FROM TEXAS

Several of the students from the summer camp joined Taekwondo and are excelling with it.

They are now integrated

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# HCU HERO: MASON FROM TEXAS

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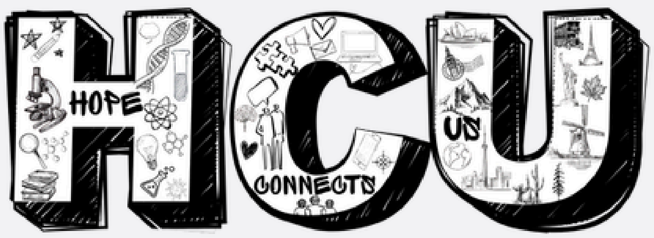
# Hope Connects Us Around the World



HCU NETWORK AMERICA

Syntis Bio

## 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.*

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 $5,780 for our Education & Outreach programs!**

# Hope Connects Us Around the World

# HCU

## Raffle Winners

Syntis Bio

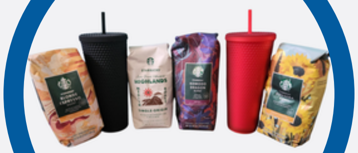
HCU NETWORK AMERICA

**50/50 RAFFLE NANCY ELLIS**

**HOLLY FULINITI**

**PEGGY SESSIONS**

**MEGHAN FIDO**



**HOLLY FULGINITI**

**JUNE ANDERSON**

**ANDY ROSE**

**LIZ CARTER**

# 2026 HCU HEROES

## RACE FOR RESEARCH

Cystadane; the last FDA approved drug for the homocystinurias was approved in 1996. That was 30 years ago!

Currently only one clinical trial 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

https://bit.ly/hcurace26

## Pricing

- Register by July 19 to receive a medal
- Register by August 24 to receive a shirt
- Early bird pricing - $30
- After July 13 - $35

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.

# It's A FAMILY AFFAIR

*In The City of Brotherly Love*

**July 10-12, 2026**
*Philadelphia, PA*

### COOKING DEMO
For Teens and Young Adults

### THE FIRST FAMILY FUN DAY!

### UPDATES ON CURRENT RESEARCH, TRIALS AND HAPPENINGS
Stay informed. Stay inspired.

### DEDICATED SCIENCE AND FAMILY TRACKS
Something for everyone!

### A CONFERENCE FOR EVERY MEMBER OF THE FAMILY
Whether you're living with HCU, a caregiver, a friend, or a supporter—you belong here.

### ENGAGING AND NEW PANELS
Featuring Patients, Caregivers and Researchers

### DINNER, AWARDS AND CONNECTION
Celebrate, honor, and connect with our amazing community.

*Patient & Family Conference*

**REGISTER NOW**

HCU NETWORK AMERICA

Classical HCU | Cobalamin Disorders | Severe MTHFR

# It's A FAMILY AFFAIR

In The City of Brotherly Love

July 10-12, 2026

Philadelphia, PA

A conference for

**EVERY MEMBER OF THE FAMILY**

— regardless of their connection to HCU! ♥

## IMPORTANT UPCOMING DATES

Mark your calendar and don't miss these important deadlines!

### HOTEL DISCOUNT CLOSES

Tuesday,
**JUNE 17TH**

Book now to secure your discounted rate!

### LAST DAY TO REGISTER

Tuesday,
**JUNE 24TH**

Don't wait—register today to secure your spot!

*Plan ahead. Save big. Be part of it all!* ♡

**REGISTER NOW**

HCU NETWORK AMERICA

Classical HCU | Cobalamin Disorders | Severe MTHFR

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

# HCU FAMILY
## Fun Day

On **Sunday, July 12,** to wrap up our biennial **HCU Network America 2026 conference,** we are hosting a fun way for our community to bond, laugh, and spend time together away from the hotel!

Join us for **activities and lunch** at Franklin Square from **10 am to 2 pm.**
We will be providing a continuous shuttle to and from the hotel for anyone who needs to return for departure.

If you would like to join us for this unforgettable experience, **please plan your travel accordingly.**

**Carousel | Mini Golf | Yard Games**

**REGISTER NOW**

# Harmony

**Now Enrolling**

**Company:** TravereTherapeutics

**Total time in study:** About 9 months

**Goal:** To learn more about classical homocystinuria (HCU), and how safe and effective pegtibatinase may be in patients with HCU

**Phase:** This is a Phase 3 study that follows a Phase 1/Phase 2 study called COMPOSE

## Who can take part?

√ People with a diagnosis of HCU

√ People 12 to 65 years of age

***Other factors and restrictions also apply***

## What is the study drug?

√ Participants will receive pegtibatinase or placebo (an inactive medication)

## Why participate in the study?

√ You may helpotherswith HCU

√ You will help contribute to new research and further understanding of HCU

√ You will receive additional monitoring and testing

Study locations include: United States, Europe, the Middle East, Asia Pacific, and South America.

70

About70people with HCU will take part in the study.

Payment for time and travel may be available to participants and their caregivers.

## Periods during the study

| SCREENING PERIOD | PRE-TREATMENT PERIOD | STUDY TREATMENT PERIOD | FOLLOW-UP PERIOD |
|---|---|---|---|
| To check if the right study is for you | To understand your current diet and how to keep it stable during the study | To receive pegtibatinase or the placebo | To check your health after the Study Treatment Period |

After completing the **HARMONY Study**, it may be possible for participants to join a long-term follow-on study and continue or start receiving pegtibatinase.

**To find out more, please contact:**
***MedInfo@travere.com***

For more information, please scan the QR code or visit:
***www.hcuconnection.com***

TRAVERE THERAPEUTICS

MA-PE-25-0038. V2.0_NOV2025.EN_US

# HARMONY GLOBAL CLINICAL TRIAL FAQ

| | |
|---|---|
| Q | What is pegtibatinase, and how is it administered? |
| A | Pegtibatinase is an investigational study drug being evaluated to help reduce homocysteine levels in people with classical homocystinuria (HCU). It is given as a subcutaneous injection (under the skin) by a healthcare provider, typically twice per week. |
| Q | How many participants are needed for the study? |
| A | Approximately 70 participants are expected to enroll globally across multiple study centers. |
| Q | What baseline homocysteine level is required? |
| A | Participants must have a baseline total homocysteine level of 50 µmol/L or higher. |
| Q | I am in a different country. Can I participate? |
| A | The study is being conducted at multiple sites across the United States, Europe, Asia-Pacific, and the Middle East. Participation depends on whether there is an active study center in your country and whether you meet eligibility criteria. Participation across countries cannot be accommodated. |
| Q | Can you maintain your normal treatment? |
| A | Yes. Participants are expected to continue their standard of care, including medications, supplements, and dietary management, without changes during the study. |
| Q | Will patients know their levels during the trial? |
| A | During the blinded phase of the study (HARMONY), homocysteine (tHcy) and methionine levels will not be shared with patients, the study doctor, or the primary physician. This helps ensure unbiased study results. Once participants transition to the open-label extension (ENSEMBLE), levels will be shared after an initial period. |

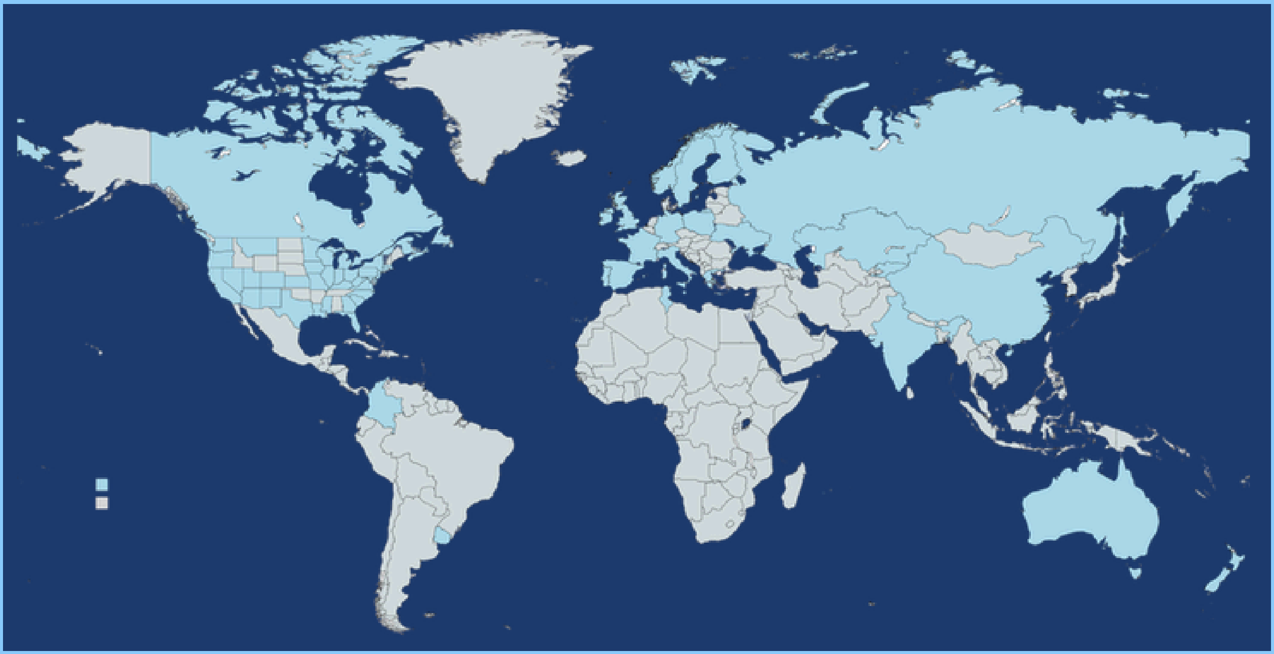
| | |
|---|---|
| Q | I work or my child is in school. Can we still participate in the study? |
| A | Yes. The study includes both clinic and home visits, and the study team will work with families to schedule visits in a way that minimizes disruption to work or school whenever possible. |
| Q | Do you have to pay to participate? |
| A | No. The study sponsor will cover the cost of the study drug and all study-related procedures, tests, and visits including travel and meals. You or your insurance will remain responsible for routine medical care that is not part of the study. |
| Q | What are the potential risks? |
| A | As with any clinical study, there are risks. Common risks include injection site reactions such as pain, redness, or swelling. There is also a potential for allergic reactions, which in rare cases may be serious. Other risks may include infection at the injection site or changes in methionine levels. Because pegtibatinase is investigational, some risks may not yet be known. Study procedures such as blood draws may also cause mild discomfort. |
| Q | What questions should I ask my doctor? |
| A | You should discuss whether this study is appropriate for you based on your medical history, current treatment, and overall health. It is also important to review potential risks, study requirements, and how participation may fit into your daily life. Your primary physician can help you make an informed decision. |
| Q | Is there a deadline to decide whether to participate? |
| A | Clinical trials do have a set end date for screening, which for HARMONY is expected to be around October of this year. Once the screening period has closed, the study won't be able to include any other participants. |
| Q | Where can I learn more about the study? |
| A | You can visit HCUconnection.com, clinicaltrials.gov or email MedInfo@travere.com. |

# HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

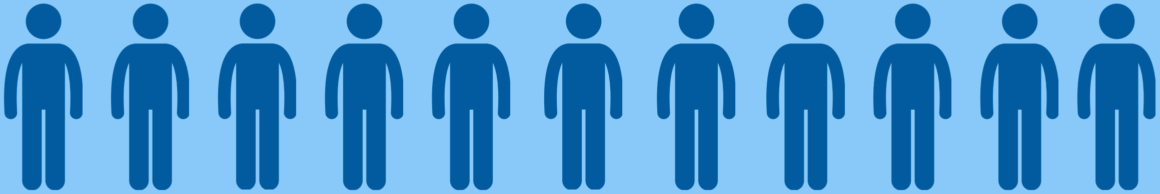
POWERED BY RAREX

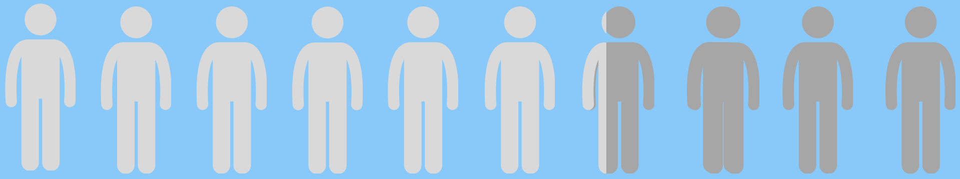
# 127 PATIENTS

# 24 COUNTRIES AND 37 US STATES



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





Only 82 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

Research
Roundup

# Identification of Variants of the MTR Gene in Patients with the cblG Inborn Error of Cobalamin Metabolism Diagnosed by Somatic Cell Complementation Analysis

David Watkins, Caitlin Zacharias, Kyana Arbabian-Urquilla, Krithika Ragupathi, Brian M Gilfix, Daniel Jimenez, Natascia Anastasio, Jean-Baptiste Rivière, David S Rosenblatt

The study confirms that changes (variants) in a gene called MTR are responsible for cblG.
The MTR gene provides instructions for making an enzyme called methionine synthase. This enzyme:

- Uses vitamin B12
- Helps convert one substance (homocysteine) into another important building block (methionine)
- Supports normal brain function, growth, and cell repair

When the MTR gene does not work correctly, this process is disrupted, leading to the biochemical and clinical problems seen in cblG.

## What did the researchers do?

The researchers studied patients who were already known to have cblG based on cell-based laboratory testing (called somatic cell complementation analysis), an older but very reliable diagnostic method.

They then:

- Examined the patients' DNA
- Identified specific genetic changes (variants) in the MTR gene
  - Identified 24 new variants, bringing the total known variants to 82
  - Reclassified 11 previous variants deemed uncertain
- Showed that these variants explain why the enzyme does not work properly

## What does this mean for patients and families?

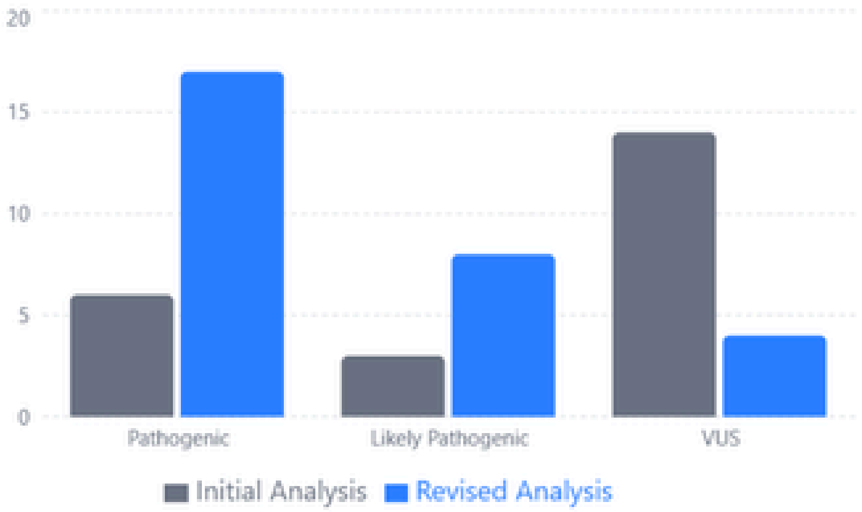
- If a person has cblG, genetic testing of the MTR gene can confirm the diagnosis.
- Families can receive genetic counseling to understand risks for future children.
- Knowing the exact genetic cause helps guide long-term care and monitoring.
- This research strengthens the medical community's understanding of cblG, which may support better care in the future

## Why is this important?

This research helps in several key ways:

- Confirms the genetic cause of cblG
- It clearly links cblG to disease-causing variants in the MTR gene.
- Improves genetic diagnosis
- Doctors can now use genetic testing to diagnose cblG more directly, without relying only on specialized cell tests that are available in few laboratories.
- Helps families understand inheritance
  - cblG is inherited in an autosomal recessive pattern, meaning:
    - Both parents are usually healthy carriers
    - Each child has a 25% chance of being affected
- Supports earlier treatment
- Researchers can now investigate the structure-function relationship of the 24 new variants

Earlier diagnosis can lead to earlier treatment (such as specific forms of vitamin B12 and other supportive therapies), which may help reduce symptoms or slow progression.



**Impact of Complementation Analysis**

How functional testing shifted variant classification severity.

This graph shows the before and after understanding of the mutations that cause cblG. Knowledge is power! With a better understanding of how these mutations affect the enzyme function, researchers can find new ways to treat it.

# 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!

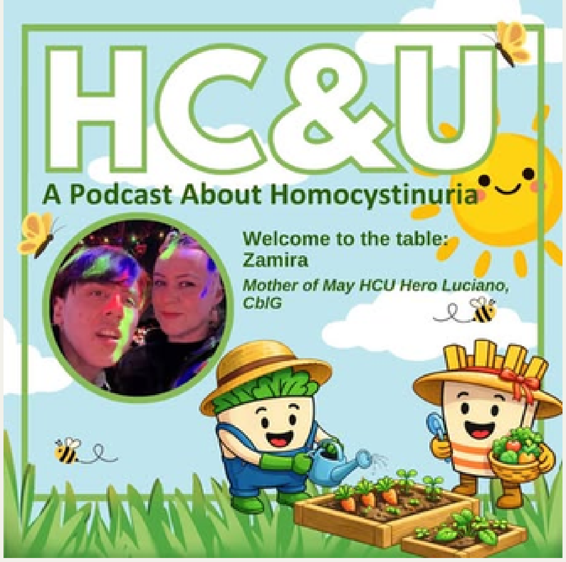
LISTEN ON Spotify

Listen on Apple Podcasts

LISTEN ON iHeartRADIO

Listen on amazon music

## The latest episode



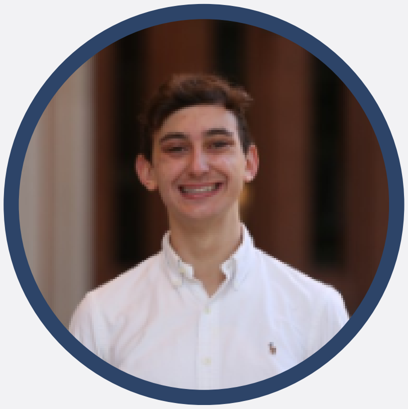
In this episode, Ben sits down with Zamira, the mother of our May HCU Hero Luciano. She shares the story of Luciano, a patient living with cblG, a rare inherited disorder that affects vitamin B12 metabolism. Luciano's journey has been marked by years of uncertainty, delayed diagnosis, and a growing list of complications—all before he finally had a name for what he was experiencing. Through his story, we explore the emotional toll of diagnostic odysseys, the everyday realities of living with a rare disease, and the resilience required to keep moving forward when answers are slow to come.

**New Series!**

# HC&U

**Bite Sized Episodes!**

## A Podcast About Homocystinuria



## Welcome to the table: Nicolo Betoni

*AN Poster and Conference*

## Welcome to the table: Kristen Skvorak

*ACMG Poster*

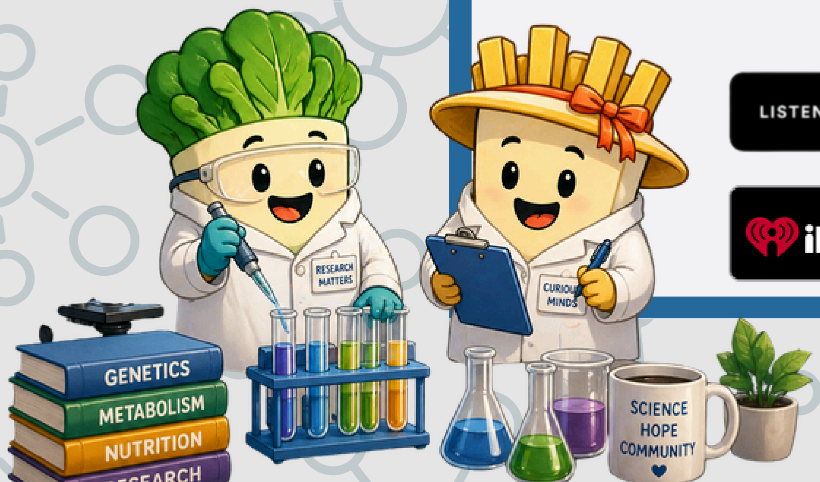
**To Listen: https://hcunetworkamerica.org/hcu-podcast/ or click below on your favorite option!**

LISTEN ON Spotify

Listen on Apple Podcasts

LISTEN ON iHeartRADIO

Listen on amazon music



# Registration Open for flok Family Camps & Adult Retreat!

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

- **Family Camp West** in Antelope, Oregon, June 4-7, 2026
- **Family Camp East** on Lake Winnipesaukee in NH, September 17-20, 2026

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.

# ILLINOIS LOW PROTEIN FAMILY CAMP

Sponsored by

NPKUA

Ajinomoto

cambrooke

BIOMARIN

pentec

Sponsored by

NUTRICIA

PTC THERAPEUTICS

Vitaflo

CYCLE

## August 7 - 9th. 2026

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.

FREE for IEM's
$5 / person
$25 / family

FUN ACTIVITIES & GAMES FOR THE WHOLE FAMILY!

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

pku

Great Oaks Retreat Center
1380 County Rd 900 N
Lacon, IL 61540

**Information:**
danaebartke@gmail.com

REGISTER HERE >

sevents.html

# Genetics Metabolism Dietitians International

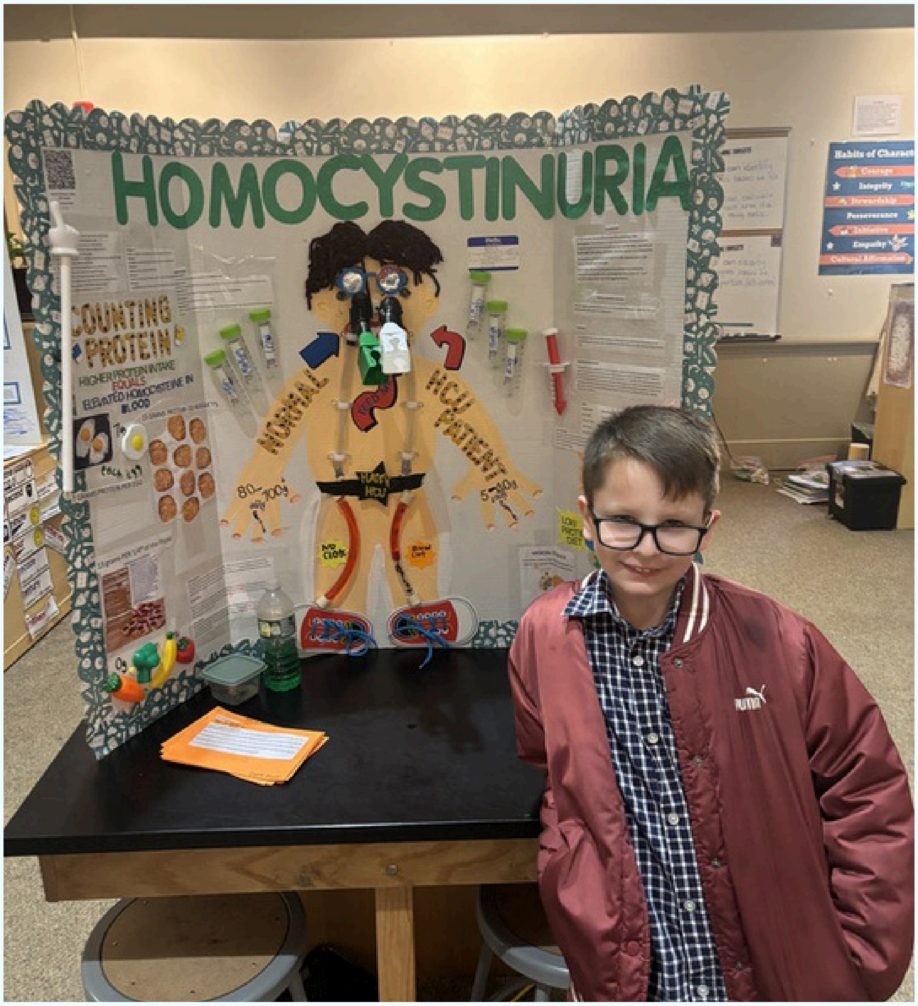
## RESEARCH GRANT PROGRAM

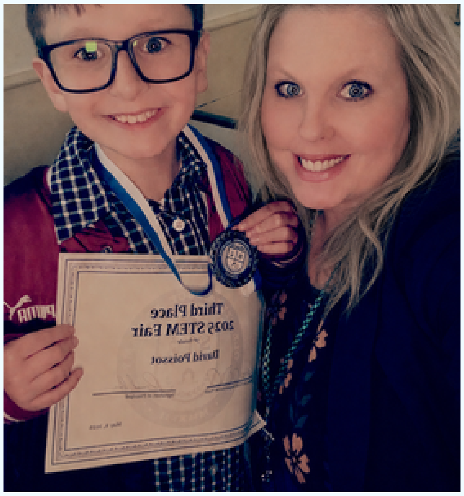
The GMDI Research Grant Program awards a $10,000 grant every two years to support research that advances nutritional management for inborn errors of metabolism. Projects must include at least one GMDI member, with preference given to clinical dietitians, junior investigators, and student researchers.

GMDI
GENETIC METABOLIC DIETITIANS
INTERNATIONAL

LEARN MORE

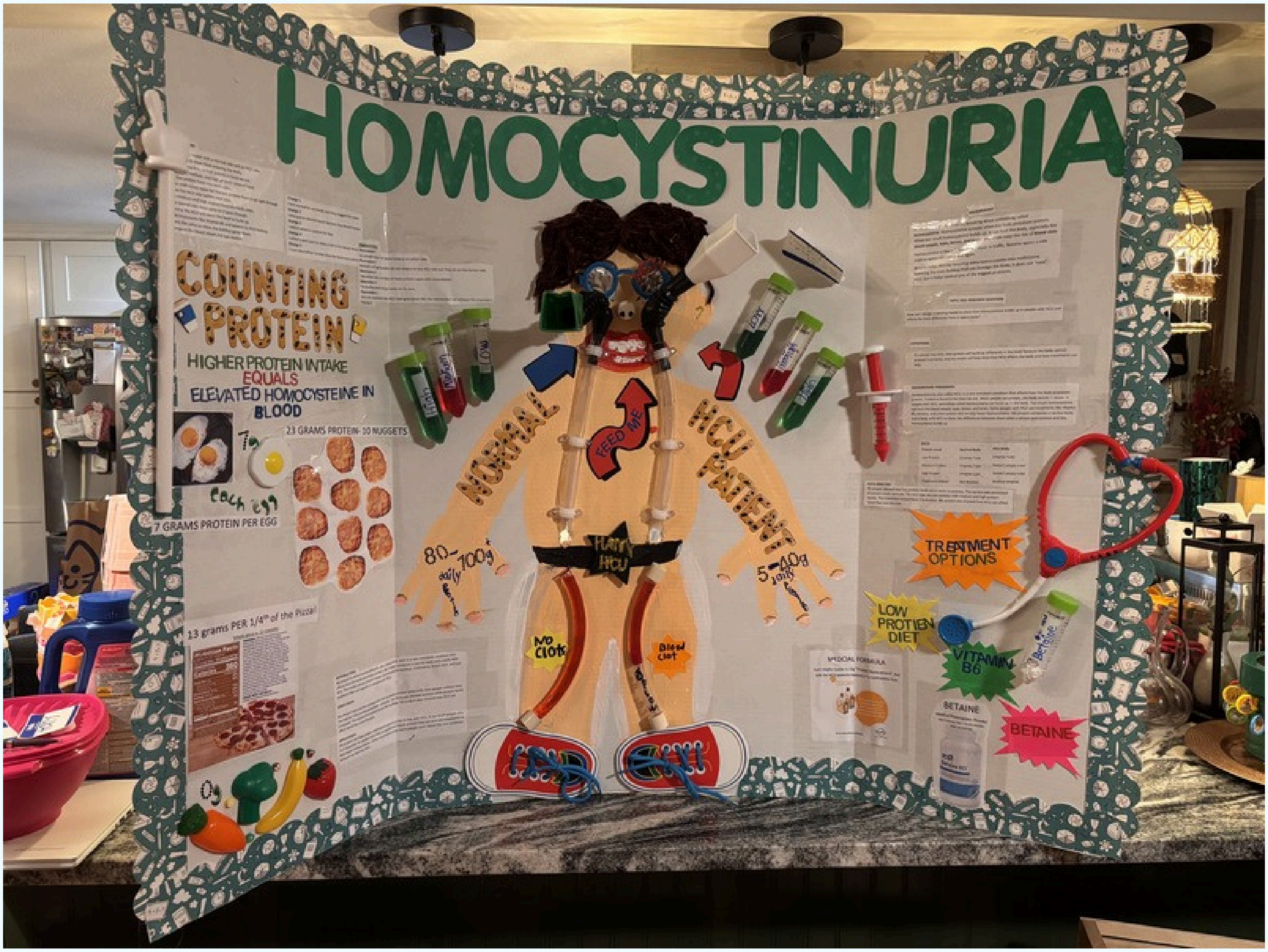
# COMMUNITY SPOTLIGHT





Meet David Poissot, an inspiring 11-year-old fifth grader at Old Sturbridge Academy in Sturbridge, Massachusetts. David has a personal connection to Homocystinuria (HCU)—his dad, Christopher, lives with Classical HCU—and this year he turned that connection into an award-winning STEM Fair project. With the support of his parents, David spent 11 weeks designing and building an interactive model that helps others understand how HCU affects the body and the importance of treatment and dietary management.

# COMMUNITY SPOTLIGHT



David’s project earned 3rd place among all fifth-grade entries and featured a working model comparing how a person with HCU processes protein versus someone without the condition. His display demonstrated key symptoms and complications of HCU, including lens dislocation and blood clots, while also explaining low-protein diets, betaine therapy, and vitamin B6 responsiveness. Through hands-on demonstrations, David showed how homocysteine can build up in the body and how treatment helps lower those levels, making a complex medical condition easier for students, teachers, and community members to understand.

HCU NETWORK AMERICA

# Meal Plan

CLICK HERE

**MONDAY**

- Buffalo Caulifower Tacos (4.9 g protein per 2 tacos)

CLICK HERE

**TUESDAY**

- Zucchini Boats (4.6 g protein per zucchini boat)

CLICK HERE

**WEDNESDAY**

- Italian Fried Mushroom Sandwich (5.2 g protein per sandwich)

CLICK HERE

**THURSDAY**

- Southwest Portobello Mushroom Sandwich (5 g protein per 1 sandwich)

CLICK HERE

**FRIDAY**

- CFL Jackfruit Kabobs (2 g protein per skewer)

CLICK HERE

## BREAKFAST

- So Delicious Yogurt w/ Berries
- Avocado Toast
- Katz Gluten Free Toaster Pastries
- Low Protein Smoothie

## SNACKS

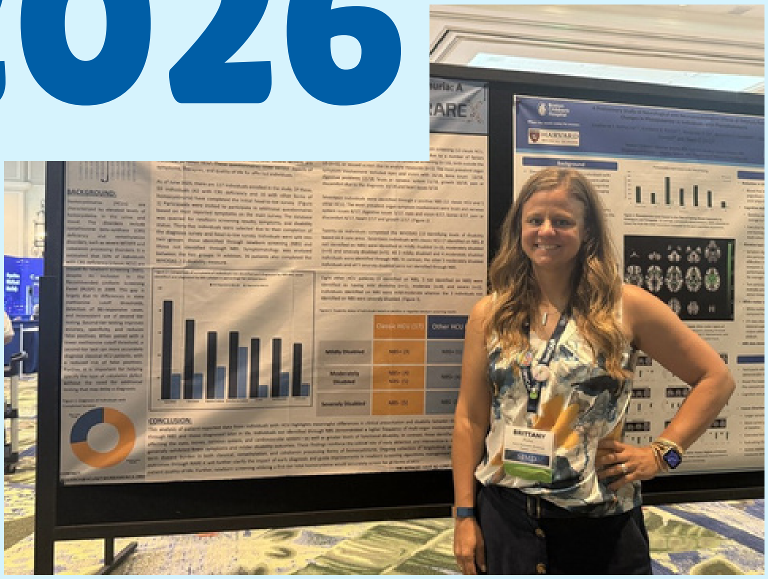
- Siete Grain Free Dip Chips with salsa
- Pirate Booty
- Absolutely Gluten Free Everything Flatbread with Field Roast Original Chao Cheese

## NOTES

- All items and recipes are hyperlinked.
- All breakfast are 2 g or less
- All snacks are 1 g or less
- **Meals should be used for lunches and dinner to reduce time spent cooking.**
- Please verify all information using the flok app.

# MAY EVENT RECAP

# SIMD 2026



We’re grateful for the opportunity to connect with members of the rare disease and metabolic disorder community, share resources, and continue conversations surrounding homocystinurias research and advocacy.

This year also included the presentation of the poster: “Patient Reported Clinical Features in Individuals with Homocystinuria: A Comparison of Newborn Screening Results.”

Thank you to everyone who stopped by, connected with us, and continues supporting progress in the HCU community. 🌍✨

# CUSTOMIZE YOUR KIT FOR FREE!

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_!

# Contact Register

## What?

- **What** is it?
  - A secure private survey for individuals or families affected by Homocystinuria
- **What** will I share?
  - Patient's birthdate, gender, exact diagnosis, and how they were diagnosed
- **What** will my info be used for?
  - Confidential and will not be shared unless we have permission
  - Helps HCUNA achieve our goals

## Why?

- **Why** should I join?
  - Able to find other families and patients in your state and request contact information
  - Access to exclusive materials (ex: we may have a webinar that a presenter doesn't want to share publicly but is okay sharing with just our community)

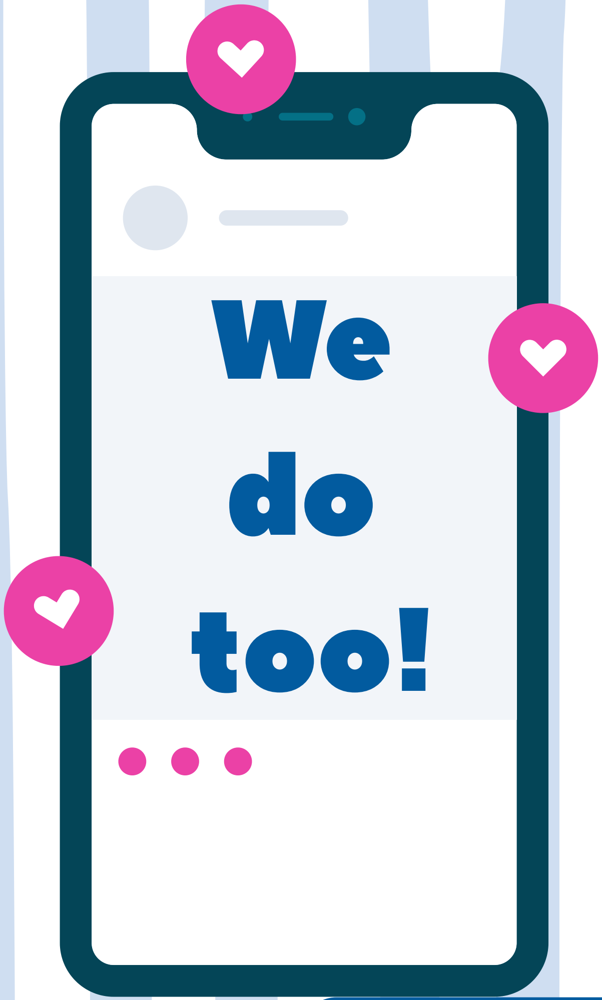
## How?

- **How** do I participate?
  - The form takes 3-5 minutes to complete
  - Visit our website and click on "contact register" tab or...

**Click Here**

## Follow Us!

# Do you have social media??



## Follow us

@hcu_network_america

HCU Network America

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