## The HCU Herald

Peruturing...

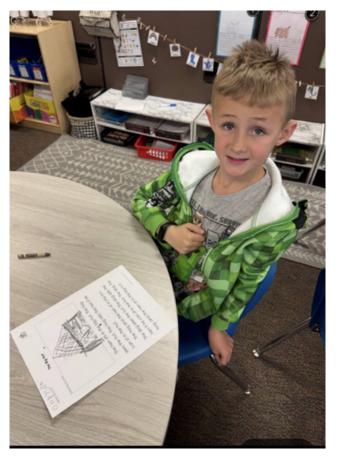


All things Homocystinuria: patient stories, resources, research, events and more!





It's been a few years since we updated the community on Grayson's progress. I like to think this is a 'where is he now' story! We previously shared our journey to a diagnosis which was long and had many trials and grief. As a family, we have been learning for the last 6 years how to live and cope with his diagnosis and make the best out of our circumstances. One of our goals is to help him understand his diagnosis, normalize it, and make sure he doesn't feel different than other kids his age.



The transition to kindergarten last year was difficult...for me, not him. It was the first time someone else would be giving him his medication outside of our immediate family. It took a lot of patience and trust. I am so grateful to his school because they did fantastic!

He didn't have any issues the entire year taking his medication. He got into a routine. After finishing his lunch, he would walk to the front office and take his medication. By the time he finished, his class was usually lining up and walking back to the room, so he would just jump back in line. Most of the class had no idea he was even gone or that he had to take daily medication. Grayson prefers it that way; he doesn't like people knowing and prefers to just take it and get back to life. He started kindergarten with an IEP for speech services and halfway through the year had met all his goals and was discharged from services. He completed the rest of the year without any support and excelled! He made lots of friends, learned how to read, write and sit still (although that skill is mostly reserved for school, not home).

Grayson is now 6 years old and about to start first grade at our local elementary school. He has spent the summer doing all the activities he loves; riding his bike, swimming at the pool, paddleboarding, playing at the park and hanging out with friends.





His big accomplishment this summer was participating in the local kids' triathlon with his big sister Riley. He swam 25 meters, rode his bike ½ mile and ran ½ mile. He's super excited for this year's HCU Race for Research because it is wizard-themed, and he is a huge fan of Harry Potter. It's been a really great experience for our family to participate together and raise funds for research.

He also played tee ball this summer and liked hitting the ball way more than standing in the outfield waiting to catch a ball. We often had to tell him to stop playing in the dirt as a ball rolled right by him. His goofy personality has defiantly emerged more recently, and we love his carefree, fun attitude.









He loves to elicit laughs from his sisters and can often be seen making silly faces, dancing like a monkey or singing songs loudly to himself. There are certainly still struggles which snap me back to reality and help me remember just how grateful I am for the HCU community.

I think the biggest support since he was born has been attending the in-person family conferences. We have attended two conferences, and they have been life-changing for our whole family. Grayson was able to meet and make friends with kids just like him.







Our daughters have met other siblings and had their feelings validated and seen. It was hard for our two girls when Grayson was born because a lot of our time and energy went to his management and treatment. They didn't complain because they love their brother and wanted the best for him, but at the same time I know they felt ignored. Robert and I were able to connect with other parents and share our struggles and successes. It's been so exciting seeing how our community is growing and all the improvements in research. We will for sure be at the family conference in 2026 and can't wait to see all our

friends and make new ones!

Grayson has been in play therapy since he was 2 years old with a clinic that specializes in working with kids who have chronic health conditions. They have worked with him to help him understand his medical



needs and the importance of taking care of himself. It has also been a great place for him to express himself and his feelings.

I think the biggest lesson we have learned is to take each day as it comes. We recognize the grief that we have over many losses, but fill it with action. As a family, we openly talk about his diagnosis and advocate for a better future for him. I'm so proud of him and grateful for the purpose he has given me in my life. I see the story of his life and the journey our family has had open doors to improve the lives of other children. I owe a huge debt of gratitude to the metabolic team in Colorado who have taken such good care of him even before he was born. I truly believe the success and growth he has had is due to his early diagnosis and treatment. His doctors keep up to date on recent publications and are always willing to listen to my ideas or concerns. Sharing his story is definitely hard because it brings with it a lot of feelings; however, the positive outcomes from others hearing our journey far outweighs any negatives. I would encourage you all to share your journey and actively be a part of the HCU community.

## Share Your Story

With Us!

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



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#### JOIN OUR FUNDRAISING TEAM

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

To Join or Learn
More:
Email
Dbartke@hcunetw
orkamerica.org



#### **UPCOMING EVENTS**





#### **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

Jessica Gold, MD, PhD Northwell Health





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



Thank you to our generous sponsors













#### WHAT IS LONGITUDINAL DATA?

DATA TAKEN OVER
TIME THAT TRACKS
SUBJECTS AT
MULTIPLE POINTS



EXAMPLE:
TAKING THE SAME
SURVEY EVERY
YEAR

#### WHY IS LONGITUDINAL DATA IMPORTANT?

OBSERVE CHANGE AND PATTERN,
STUDY TRENDS,
CAUSE AND EFFECT RELATIONSHIPS,
LONG TERM IMPACTS

THIS LEADS TO ...

- BETTER UNDERSTANDING OF HCU
- SHORTEN DIAGNOSTIC ODYSEEY
- **IMPROVE MANAGEMENT GUIDELINES**
- DEVELOP NEW AND BETTER THERAPIES

  FIND A CURE!









#### IN CASE YOU MISSED IT...



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!









#### The latest episode



Ben welcomes Hannah to the table! Hannah is the mother of Olive, who was diagnosed with Homocystinuria Cobalamin G after a long and difficult diagnostic odyssey. Their journey to diagnosis in the United Kingdom is very similar to those experienced by families around the globe. HCU is grateful to our international community for continuing to share their experiences and hopes for the future.



## 2024 TEAM FEATURES

#### "HUNT" FOR RESEARCH



Team "Hunt" For Research raised **\$31,719 and totaled 537 miles** in the 2024 Race for Research.

Congratulations to Carson
Hunt and his teammates
Megan Lauvetz, Stacey
Dickerson, Abby Dickerson,
Kari McGaughey, Ethan Hunt,
Dana Hunt, Brad Clay, Darren
Hunt, Matthew Allen, and Sue
Lauvetz!

Team Butter Stick raised **\$6,577** and totaled **73.37 miles** in the 2024 Race for Research.

Congratulations to Audrey Hill, Janine Melink-Hueber, Danny Hill, Jennifer Rammel, and Bethany Bingham, all on behalf of Elijah, who has a Cobalamin E Deficiency.

#### **BUTTER STICK**







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.





#### 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



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



https://bit.ly/HCURace25





#### 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, ageappropriate 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

## COME SEE US AT

Canpku Conference

August 22-24, 2025 Calgary, Alberta

#### **PRESENTATION**

3:15 pm-3:45 pm Exploring the Rare-X- HCU Registry Speaker: Brittany Parke

#### **POSTER**

"Incidence of Neurocognitive Symptoms in Patients with Homocystinuria"







#### **UPCOMING EVENTS**





Your rare condition.

Our common fight.



Save The Date
July 10-12, 2026

HCU Network America
2026 Conference
Philadelphia, PA





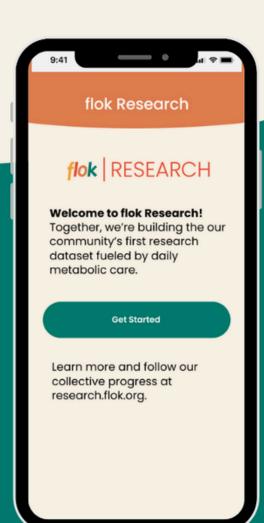
## ADULTS WITH CLASSICAL HCU



Join the 12-week Signal Sub-Study at flok Research to uncover patterns in diet, sleep, symptoms, and well-being.

#### **Now Enrolling!**

Open to flok App users age 18+ living in the US.







Learn more at research.flok.org and join flok Research via the app's main menu



#### Family Camp East | Sept 18-21 | Tuftonboro, NH

- Adventure & activities for all ages
- Low-pro meals
- FREE for anyone with Classical HCU
- Register by August 17
- Details at flok.org/camp









## Registration is now open for the 2025 Newborn Screening Bootcamp

Co-hosted by the <u>EveryLife Foundation for Rare Diseases</u> and <u>Expecting Health</u>, this virtual Bootcamp offers participants the chance to learn from experts and patient advocates actively involved in the newborn screening process.

For more than 50 years, every newborn in the U.S. has been screened for a range of serious and potentially fatal conditions using a dried blood spot collected via heel prick. Each year, approximately 14,000 babies and their families benefit from early detection and access to life-saving treatments thanks to newborn screening.

This event is designed to foster meaningful discussion and share key developments across the newborn screening ecosystem.

Who can participate? Anyone interested in learning more about newborn screening is welcome to attend.

The 2025 Bootcamp will be held virtually on September 17, 2025 12:00pm – 4:00pm ET.





#### **GET YOUR KIT**



## Our FREE Customizable Kits are here!



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 - <a href="https://www.surveymonkey.com/r/HCUKitSurvey">https://www.surveymonkey.com/r/HCUKitSurvey</a>

\*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!

# Important Message For those taking Vitaflo HCU Gel

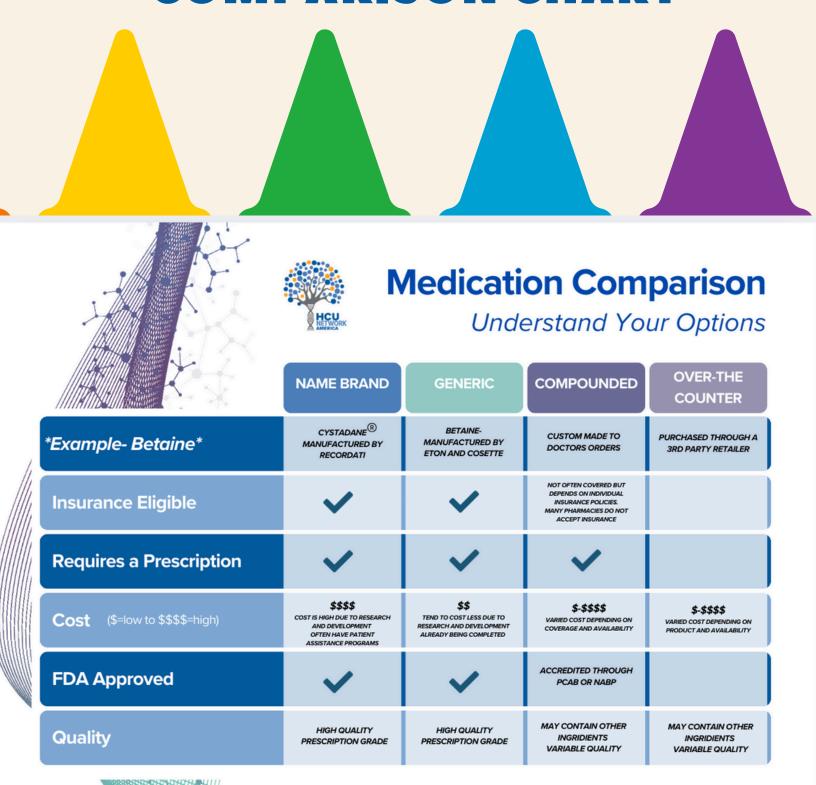
Vitaflo will be discontinuing this formula. No new product will be made for the HCU Vitaflo Gel product, and they project that they will have a supply for the coming months, depending on the demand.

If you take this formula, please speak with your healthcare provider to find a suitable alternative.





#### MEDICATION COMPARISON CHART





## RESEARCH OPPORTUNITY

The Children's Hospital Zurich is developing a new questionnaire to assess health-related quality of life in people with Classical Homocystinuria (HCU)- and they need your input.

They are looking for:

- Children's & tens 8-18 with Classical HCU
- Their parents
- · Adults with Classical HCU

Your participation will directly support better care and evolution of new therapies.



Helene.Werner@kispi.uzh.ch martina.huemer@kispi.uzh.ch







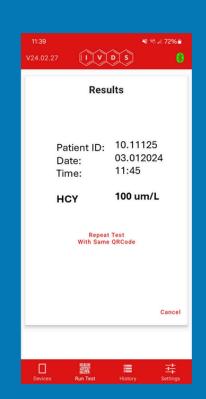
#### **BREAKING NEWS**

In Vitro Diagnostic Solutions (IVDS)
has received funding for their Phase I
grant award from the National
Institute of Health for the Hcy Now.

The Hcy Now is "a point of care device for determination of total Homocystiene (Hcy)"

TO LEARN
MORE ABOUT
IVDS







#### BACK TO SCHOOL RESOURCES



#### **Navigating Accomodations**

\* también disponible en español



Parent Handbook for Special Education Services

#### Low Protein Lunch resources

Cambrooke school lunch program & more

#### College Transition



Customizable "All About Me" pamphlet

#### Homemade Low Protein No Crust Sandwiches





#### Ingredients/Tools:

- Two slices of low protein bread of choice
- Cookie butter (brand of choice)
- Jelly (flavor of choice)
- Sandwich cutter (shape of choice)

#### Instructions:

- 1. Make your sandwich as normal
- 2. Use your sandwich cutter to remove the crusts

#### Tips:

- 1.Use a softer bread and avoid dry brittle bread
- 2.Use cookie butter on both sides of the bread
- 3. Fill less than you would think to
- 4. Freeze if you are making multiple for the week Free

The protein count varies depending on the brand you use. Do your own research before trying this.



#### Now includes funding assistance for medical formula AND low protein foods!



#### CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

#### What is the purpose of this program?

Having a rare disease is difficult. Adding in the complex care required to treat or manage that disease and figuring out how to pay for it makes a rare diagnosis even harder.

NORD's Classical HCU Medical Assistance Program offers eligible individuals diagnosed with Classical Homocystinuria financial support to pay for the low protein foods and physician prescribed medical formulas necessary in managing this HCU diagnosis.

NORD's HCU Medical Assistance Program opened thanks to a generous donation from the HCU Network America.



NORD, a 501(c)(3) organization, is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. NORD, along with its more than 300 patient organization members, is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services.

**MEDICAL** 

ASSISTANCE

NORD was founded by families struggling to obtain access to treatments and whose advocacy for change led to the passage of the Orphan Drug Act in 1983. NORD assists eligible patients (those with medical and financial needs) in affording the treatments and medical services their healthcare professionals have prescribed.



#### Who is eligible to apply?

This program is designed to help patients who:

- · Have a diagnosis of Classical Homocystinuria.
- Are a United States citizen or U.S. resident of six (6) months or greater with evidence of residency such as a utility bill showing the patient's name and address.
- Meet the program's financial eligibility criteria.



#### What is the application process?

Patients may be referred to the program by their health care provider, their case managers, or they may self-refer.

A NORD Patient Services Representative will guide the applicant through the application process and verify eligibility for inclusion in the HCU Program.

Awards are based on meeting eligibility criteria, funding availability, and are made on a first-come, first serve basis.

Alone we are rare. Together we are strong.®

#### How do I get more information and apply?

Contact NORD's Classical HCU Medical Assistance Program

Monday-Thursday 8:30am – 7:00pm ET Friday 8:30 am – 6:00pm ET



203-616-4327



203-635-4163



hcu@rarediseases.org



US MAIL to: NORD Attention: HCU Program 55 Kenosia Avenue Danbury, CT 06810



- The Classical HCU Medical Assistance Program assists eligible individuals with out-of-pocket costs to purchase low protein foods and physician prescribed medical formulas. Individuals approved for assistance in this program will be issued a PEX card. The PEX card is a prepaid expense card to be used for the purchase of low protein foods or physician prescribed medical formulas only.
- Upon receipt of the card, the cardholder will contact NORD to request card activation. The card will be funded based on program award caps set for the program (this cap will be discussed with individuals upon enrollment in the program...
  - > It is necessary for the individual to submit receipts on a monthly basis evidencing card utilization for the purchase of low protein foods or medical formulas for the previous month.
  - > Funds will not be added to the card until the previous month's receipts have been received by NORD.
  - > The card may only be utilized for the purchase of low protein foods up to the monthly program limit.

#### Once a patient is accepted into the assistance program(s) how long are they eligible?

Awards are issued for a calendar year.

Patients are encouraged to reapply annually if continued assistance is needed.



#### What happens if an applicant does not meet the criteria of the Electronic Income Verification?

The NORD Patient Services Representative will offer to e-mail, fax, or mail the brief program application and disclosure forms to the patient. The applicant may then complete the application, sign the disclosure form, provide the appropriate financial documentation to verify financial need, and return them via fax, email, or USPS mail.

#### How does NORD demonstrate compliance with regulations required of charities?

- NORD independently designs its patient assistance programs based on the needs of specific patient communities.
- No pharmaceutical company or donor controls or influences our programs.
- Our patient assistance decisions are based on consistently applied financial eligibility criteria and diagnosis only.
- Patients have their choice of health care provider, treatment and treatment location, and can make changes at any time.
- Patients' privacy and well-being are priorities at NORD. We
  do not share or provide patient names or data with donors,
  nor do we disclose or identify donors to patients. Patients
  are able to make the choices that are best for them
  because NORD's assistance covers all FDA-approved
  products available for a diagnosis. Our programs also help
  with more than medication: patients can use their funds to
  pay for other physician prescribed services related to their
  diagnosis, such as laboratory and diagnostic testing,
  physical and occupational therapy, durable medical and
  adaptive equipment, and travel to medical appointments.

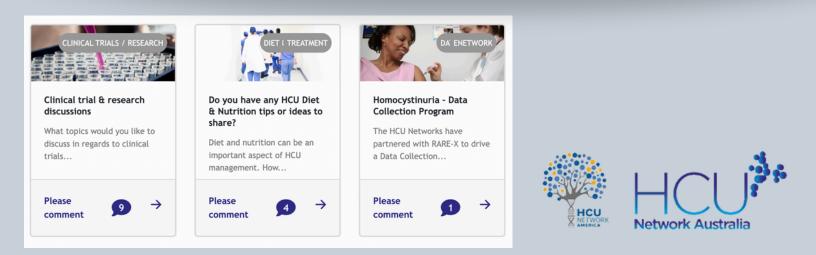


#### Welcome to the HCU eNetwork



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.



Join the conversation!

https://hcuenetwork.org/

- 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 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)

## What?

## Contact Register

## 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!

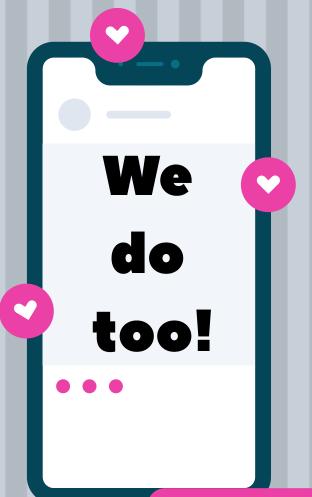








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