The HCU Herald

Featuring...

HCU Hero
Ellie from Pennsylvania

March 2024

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

HCU HERO:
ELLIE FROM PENNSYLVANIA

UPDATE

5-year update Edition!

In 2018, when Ellie was 6 months old, her family shared her HCU Hero story with us for the first time.
You can read her original story here.

Keep reading to find out how Ellie is doing today!

As Ellie approaches her 6th birthday, we reflect on the incredible journey she's had since those challenging days at the hospital five years ago. She's grown into a strong and fearless girl, loving dance, gymnastics, and now aerial acrobatics, with determination. She's earned the nickname "cannon arm" in softball, showcasing her strength and resilience.

As we face occasional hurdles with her HCU levels or medications, we remain grateful for the advancements in newborn screening and genetics research.

Navigating new challenges, Ellie sometimes faces days when food choices are tough. Whether it's dealing with not eating or frustration over dietary restrictions, these moments can be challenging. Going to friends' houses and

HCU HERO: ELLIE FROM PENNSYLVANIA

managing her diet at school introduce additional hurdles. Despite this, Ellie's resilience continues to shine, and we're committed to supporting her through each step of this journey. Together, we'll find solutions and keep embracing the strength that defines her.

We are so proud of Ellie and who she's becoming. She has taken the challenges of HCU and used it to make her more determined and a leader in most situations.

Keep reading for a special feature:

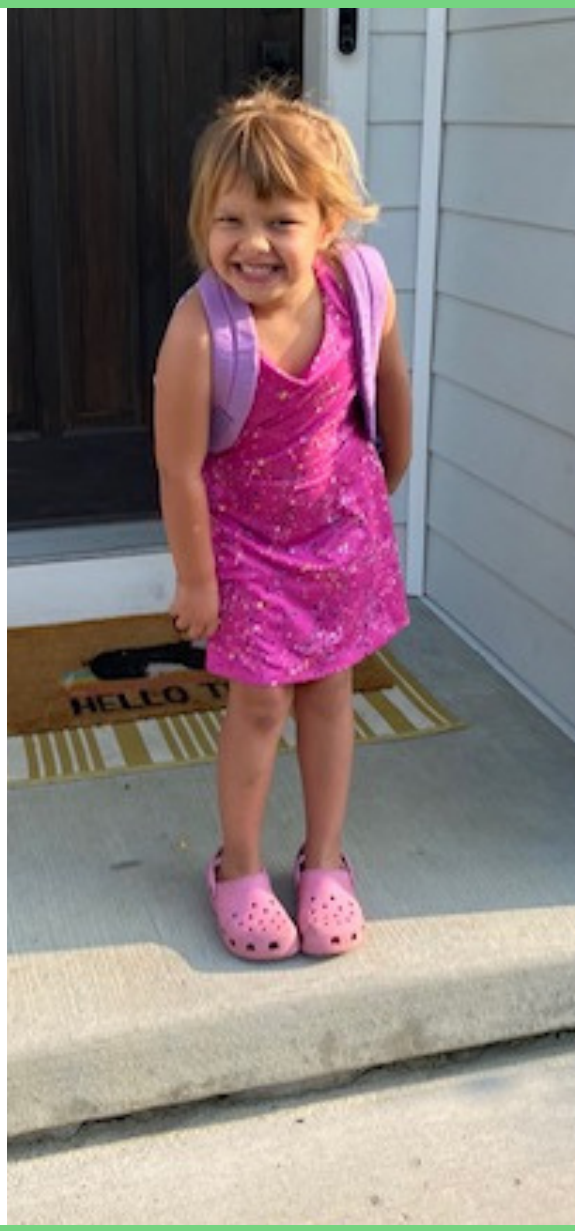
Interview with Ellie's mom!

INTERVIEW WITH ELLIE'S MOM

We asked Ellie's mom, Jeannie, some questions to learn a little more about Ellie, her experience as a kindergartener, with the low protein diet and formula, as well as their family's hopes for the future. Here's what we found out!

What is Ellie's current protein allotment? What are some of her favorite foods, or foods that work well within her allotment?

Ellie is at 150 mg of methionine- or about 8.5 g of protein. This was a decrease from last year when she could have 9.5 g of protein. It doesn't seem like much, but the 1 g difference changed most of our go-to meals for her. Currently she has been asking for avocado toast using gluten free bread. We like to pair it with some fruit, like apples or strawberries, to have a more complete meal. Her go-to lunch is macaroni and cheese using gluten-free pasta and a variety of cheeses. The trick is to add some vegan cream cheese to make it creamy! We like to add peas or green beans to give her more to eat at a lower-protein level. Gluten-free pretzels and vegan french onion dip are a must right now. We can give her a full serving of pretzels, dip, plus some cucumbers, and create a whole lunch for about 1 g of protein.



INTERVIEW WITH ELLIE'S MOM

How does Ellie do with the formula? Any tips/ideas that you guys have found to work well at helping her get her formula in?

Ellie goes through phases with formula. Sometimes she will drink it quickly, and sometimes it feels like she is taking in drops at a time. Currently, she is drinking the formula just fine but needs constant reminders to "take a sip" - she is 6 and easily sidetracked! It takes her about 10 minutes to finish her allotment, 3 times per day. When Ellie is going through a phase of not wanting to drink it, it's typically because she wants control over the situation. We will give her some control by letting her pick which cup she wants to use, or what she wants to do while drinking it. When it gets really tough, we will wait for her to want something and I will respond by telling her "I will get you what you want, as soon as your drink is finished." Luckily these things have worked for us, and the phases don't last too long.

What grade is Ellie in? What has her school experience been like?

Ellie is in kindergarten this year. We pack her lunch, and she takes her lunch time formula to the nurse every day. There have been a few times that her class has had snacks, but we typically know beforehand and will send a special snack with her. The few times that they have had a last-minute treat, her teacher has texted or called me with the information, and I let him know if she can have it. A few times she couldn't have what they were providing, so they got an apple from the lunchroom for her to participate.

Ellie takes full ownership of her bottle time. At 11:07 she tells the teacher it's time for her to go to the nurse, and she goes. Her teacher mentioned he doesn't

INTERVIEW WITH ELLIE'S MOM

often even get to his reminder; she usually reminds him first. The biggest issue we are having is her eating at lunch. She tends to feel full from her bottle or is sidetracked and leaves half of her lunch uneaten. It makes it difficult to track and she is often hungry as soon as she gets home. These are all things we are making adjustments for currently to help the situation.

What are your/your family's hopes for potential future treatments or developments for HCU?

I would love an enzyme replacement therapy that requires minimal injections or disruptions to her life. I want her to travel and experience life without being chained to scales, formula, and medicine. Not only are the medicines expensive, but the diet is too. Gluten-free and fresh fruit/veggies are always putting us at a higher grocery budget. I worry that as she gets older, she won't be able to pursue her dreams because she'll make choices that won't allow her to afford these things more readily, again being chained to something she doesn't want. If there were a medication that would allow her to be more independent, that would be our hope.

To read Ellie's 5-year-update story on our website, visit:
https://hcunetworkamerica.org/ellie/

We need Patient Stories!

BY SHARING YOUR STORY, YOU CAN....

- Spread awareness
- Help others to feel connected
- Share insight beyond our patient community of what it's like to live with homocystinuria
- Provide hope to other families

Ready to share your story?

Send us a message on social media or email us!

info@hcunetworkamerica.org

HCU NETWORK AMERICA

JOIN OUR FUNDRAISING TEAM

We are looking for new community members to join our fundraising team!

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

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

To join, email Dbartke@hcunetworkamerica.org

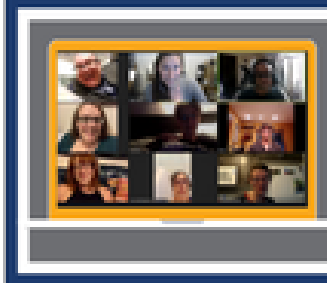
UPCOMING EVENTS

Our teen (10-22) meetups are an opportunity for teens with any of the homocystinurias to connect, learn, and share experiences.

Register:

https://bit.ly/teen-meetup

Our virtual meetups are an opportunity for patients & caregivers with any of the homocystinurias to connect, learn, and share experiences. We will also share important community updates!



Register:

https://bit.ly/hcu-meetup

Registration:

TICKETSIGNUP.IO/TICKETEVENT/HCUCONFERENCE

MOVING MOUNTAINS

Classical HCU | Cobalamin Disorders | Severe MTHFR

Patient-Family Conference

Aurora, Colorado
June 29-30, 2024

HCU NETWORK AMERICA

- KEYNOTE FOCUSED ON MENTAL HEALTH
- AGING WITH THE HOMOCYSTINURIAS PANEL
- BREAKOUT SESSIONS
- DIAGNOSTIC TOOLS AND NEWBORN SCREENING
- RESEARCH UPDATES
- HCU HERO AWARD RECEPTION

Incase 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!

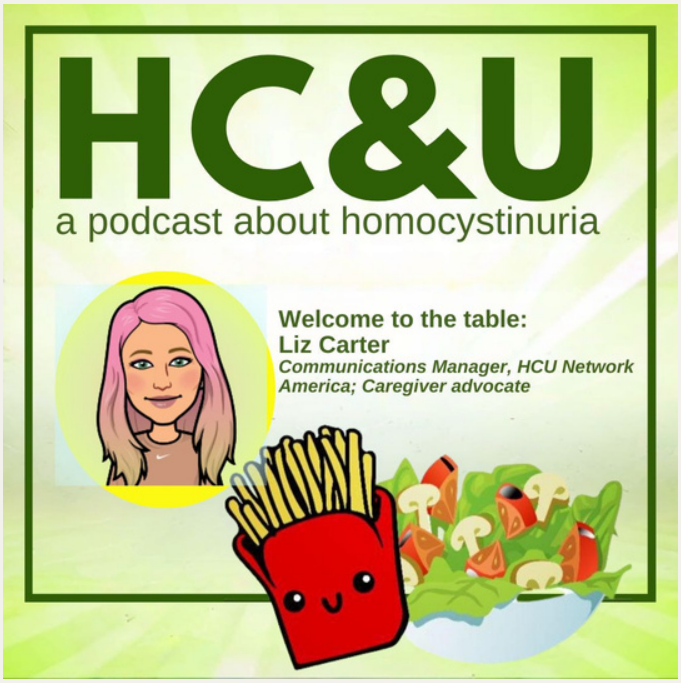
Welcome to the HC&U Podcast! We are Ben and Lindsey, your hosts. We are so excited to be starting this as extra resources for the Homocystinuria community. We hope you like our content!

To Listen:
https://hcunetworkamerica.org/hcu-podcast/
or catch it on Spotify or Apple Podcasts!

LISTEN ON Spotify

Listen on Apple Podcasts

The latest episode



In this episode, Ben welcomes Liz Carter, mom of 7-year-old Elliott, who lives with Classical Homocystinuria. Ben & Liz chat a bit about Elliott's journey and how his being missed at newborn screening led Liz down a unique advocacy path. Liz also shares some ways that anyone looking to get involved with advocacy can get plugged in!

Sign our NBS Screening Petition!

In this petition, we call for state newborn screening labs to **revise screening protocols for Classical Homocystinuria** to *ensure fewer false negative screening results and delayed diagnoses.*

Click here to sign the petition!

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

Diagnosis Survey

Take the survey now at: homocystinuria.rare-x.org

Newborn Screening & the Homocystinurias

The approximate number of babies with Classical HCU that are missed at Newborn Screening (however, some reports suggest up to 80% are missed.)

The number of states that screen for classical homocystinuria. Classical HCU was added to the RUSP (Recommended Uniform Screening Panel) in 2007, thanks to the Newborn Screening Saves Lives Act.

Cobalamin Disorders & Severe MTHFR

Combined Cobalamin Disorders are a part of the RUSP Secondary Conditions. This means they are picked up as a result of a different disorder, typcally Methylmalonic acidemia. These condtions are Cobalamin C, F, J, K, X and TC II.

Some states set a low methionine cut off and these will flag the conditions above, but also flag Isolated Cobalamin Disorders, Cbl D, E, G & Severe MTHFR.

What's YOUR Diagnosis story?

Diagnosed at birth? Not screened? Missed? Let us know!

CANPKU+ CANPGU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

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*!

SCHOLARSHIP OPPORTUNITY!

Pursue Your Dreams through the #RAREis Scholarship Fund

Living with a rare disease means managing unique challenges, including frequent doctor visits, rigorous treatment regimens and hospitalizations, and exposure risks. While quality and duration of life continues to improve thanks to improved diagnosis and treatment approaches, individuals living with rare diseases still face disparities in achieving traditional life milestones. That's why the EveryLife Foundation for Rare Diseases established the #RAREis Scholarship Fund – to enrich the lives of adults living with rare diseases by providing support for their educational pursuits.

Thanks to the support of Amgen,* The EveryLife Foundation for Rare Diseases will provide one-time awards of **$5,000 scholarships** to **53 rare disease recipients** in 2024. *Horizon Therapeutics was acquired by Amgen in 2023.*

Who can apply?

Anyone 17 or older, who is a resident of the United States and who has been diagnosed by a physician as having any form of rare disease, regardless of treatment status.

When are applications being accepted?

March 18- April 22, 2024

What schools/universities qualify?

Applicants must plan to enroll full-time or part-time in undergraduate or graduate study at an accredited two- or four-year college, university, or vocational-technical/trade school for the Fall 2024 semester. There is no minimum amount of credit hours to be part-time. Students do not need to be pursuing an undergraduate or graduate degree.

Click here to learn more and to apply!

Scholarship opportunity!

flok The Guthrie-Koch Scholarship

- **Do you have Classical HCU?**
- **Are you a *high school senior* or *current student* pursuing an undergraduate degree or technical school?**

If you answered 'yes', you are eligible to apply for the Guthrie-Koch Scholarship Program!

The Guthrie-Koch Scholarship Program was founded in 1997 to recognize outstanding young adults with PKU pursuing higher education and provide financial support to these efforts, but has now been expanded to include young adults with Classical HCU and other metabolic disorders!

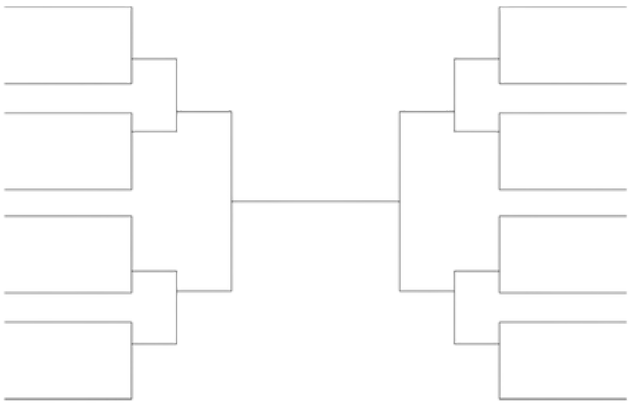
Applications for the 2024-2025 academic year are now being accepted at scholars.flok.org.

The application deadline is March 15, 2024.

HOST YOUR OWN MARCH MADNESS FUNDRAISER!

March Madness Selection *Sunday 2024 is* ***March 17!***
Start assembling your pool & raise funding for resources, tools & outreach for **HCU Network America!**

March Madness is a three week period packed full of buzzer beaters and is a sports poolers dream. But how can you keep the excitement alive when most of your members have had their brackets busted? A great option is our Madness Squares pool format, as every game of the tournament will have a winner!



How Do Madness Squares Work?
If you are familiar with Super Bowl Squares, the main idea is the same for March Madness. A 10x10 grid of boxes is setup and each row and column is given a number from 0 to 9. Just like in Super Bowl Squares, each square of the grid can be claimed by a pool member.

Winner breakdown
Each round is worth a set number of points. You can determine this on your own, but be sure to let all of the entries know before the tournament begins what the scoring system will be. (You should write the point values under each round at the top of the bracket).

Declaring a Winner
Multiply the total number of correctly picked games in each round by the points assigned to that particular round. Tally all rounds together and the person with the highest point total wins!

- For further instructions and to print your bracket, visit: https://www.printyourbrackets.com/howtomarchmadness.html
- For online tools, check out: https://www.runyourpool.com/march-madness-squares-pools.cfm

Industry news

Earlier this month we shared Synlogic's announcement to terminate their PKU study, and to no longer pursue a program for HCU. We share in deep disappointment for the PKU community, as well as our own.

While this particular program may be ending, the initiation of a Phase III trial of pegtibatinase continues to provide hope for the HCU community.

Read Synlogic's press release here

UPDATE

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

NORD®

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.

MEDICAL ASSISTANCE

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.

NORD is Here for You

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.

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.

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

What assistance does NORD provide?

- 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.

NORD®

rarediseases.org

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**

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