The HCU Herald

Feauturing...

HCU Hero

Everett & Clementine

From Colorado

September 2025

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

HCU HERO:
EVERETT AND CLEMENTINE FROM COLORADO

Clementine was born in February 2025 in Denver, Colorado. Our third baby, but first little girl, we were overjoyed to have a daughter complete our family. Clementine's big brothers, Everett (5.5 years old) and Oliver (3 years old) anxiously awaited her arrival and have always adored their little sister. The pregnancy and birth were smooth and without complications.

Clementine breastfed well from the start and began gaining weight quickly. While in the post-partum wing of the hospital, Clementine received her first newborn screening at 24 hours old, and again at

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around 2.5 weeks old at her 2-week check up at the pediatrician. Having already had 2 babies pass their newborn screens and Clementine seemingly a perfectly healthy and normal baby, we knew the drill and didn't think much of it.

Two weeks passed and nothing seemed out of the ordinary. We were settling into life with a newborn, toddler, and preschooler, and all the chaos that comes with it. On a Friday evening when Clementine was 4 weeks old, we received a call from her pediatrician.

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Clementine's newborn screen was abnormal and we needed to bring her to Children's Hospital right away for bloodwork to confirm the results. We, of course, immediately panicked and began furiously Googling what conditions were included in Colorado's newborn screening. We took her for bloodwork the following day, which revealed a homocysteine level of 153.9 mcmol/L and a methionine level 736 nmol/L. At the time, we did not know what this meant other than that the levels were WAY beyond the normal range outlined in the test results. After more furious Googling, we realized Clementine likely had a metabolic condition we had never heard of—homocystinuria.

By Monday morning, we were frantic for more information. What did this mean? Was she in

immediate danger? Was there treatment? A cure? What do we do?! That day, we were contacted by a Genetic Counselor with the Metabolic Clinic at Children's Hospital. She confirmed what we suspected – Clementine had a condition called homocystinuria and needed to be seen by the Metabolic Clinic team right away. We met with the Metabolic team the following day, who confirmed Clementine's diagnosis. We learned that our other children had a 25% chance of having HCU. But both Everett and Oliver had passed their newborn screenings without issue! Weren't they in the clear? No, they weren't.

We got the boys' blood drawn right away to test their homocysteine levels. Oliver's was 5.1 mcmol/L and in the normal range. Everett's homocysteine was an astounding 222.7 mcmol/L.

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When we saw the results, my heart plummeted into my stomach. I felt sick. How could this have happened? Everett was tested as a newborn and passed! We were devastated.

Distraught. Furious! We had missed the chance to treat Everett from birth, and now he would suffer more adverse impacts from the condition for the rest of his life.

The reason Everett was not flagged for HCU in his newborn screen is that Colorado's threshold for identifying HCU in the newborn screening process was too high when he was born.

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Between the time Everett was born (2019) and Clementine was born (2025), Colorado lowered the threshold substantially. As a result, Everett was missed in newborn screening, but Clementine was caught. Colorado is a two-screen state (1 of 10 in the US), which means babies are first screened at 24 hours old. If a baby is flagged in the first screen, the baby is re-tested again at 8-14 days of age. The two-screen process is very important because it allows a very low methionine cut off since the babies initially flagged will be re-screened and will trigger an abnormal finding only if the second screening shows out-of-range methionine levels.

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This is a crucial aspect of the newborn screen process for HCU, as newborn babies simply don't have high enough methionine levels to meet a high threshold when they are first born.

As for Colorado, the state began checking for HCU in the newborn screen in 2006. From July 2006 to November 2021, newborns in Colorado were flagged for HCU in the first screen only if their methionine was 100 umol/L or higher.

During this period, no newborns with HCU were identified through newborn screening.

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This included Everett, whose first newborn screen measured a methionine level of 83 umol/L. Because his methionine was below the 100 umol/L threshold, his screen was deemed normal and his methionine was not measured again on his second screen. Consequently, Everett was missed by newborn screening.

In November 2021, the Colorado Department of Public Health and Environment reduced the methionine cut-off in the first newborn screen to 47 umol/L, but still no babies were flagged for HCU. CDPHE then lowered the methionine cut-off even further, to 33 umol/L, in August 2022. With this new threshold, 2 newborns were finally flagged for HCU through newborn screening.

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One of these babies was our daughter Clementine, whose first newborn screen in February 2025 showed a methionine level of 70 umol/L. Because her methionine was above the 33 umol/L threshold, Clementine was re-tested at her 2-week pediatrician appointment, which showed her methionine had climbed to a whopping 616 umol/L. It was at this point that CDPHE reached out to the Metabolic Clinic, who in turn contacted our pediatrician so that we could have Clementine undergo diagnostic bloodwork at 4 weeks old.

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Clementine is the first newborn baby in Colorado with HCU flagged through Colorado's newborn screening process. Until Clementine was born in February 2025, no babies with a methionine below 100umol/L with HCU had ever been caught in newborn screening in Colorado, including our son Everett.

While Clementine was flagged and began treatment for HCU before she was 6 weeks old, Everett was missed and, therefore, wasn't diagnosed or treated for HCU until he was 5.5 years old. Everett will bear the consequences of the later diagnosis his whole life.

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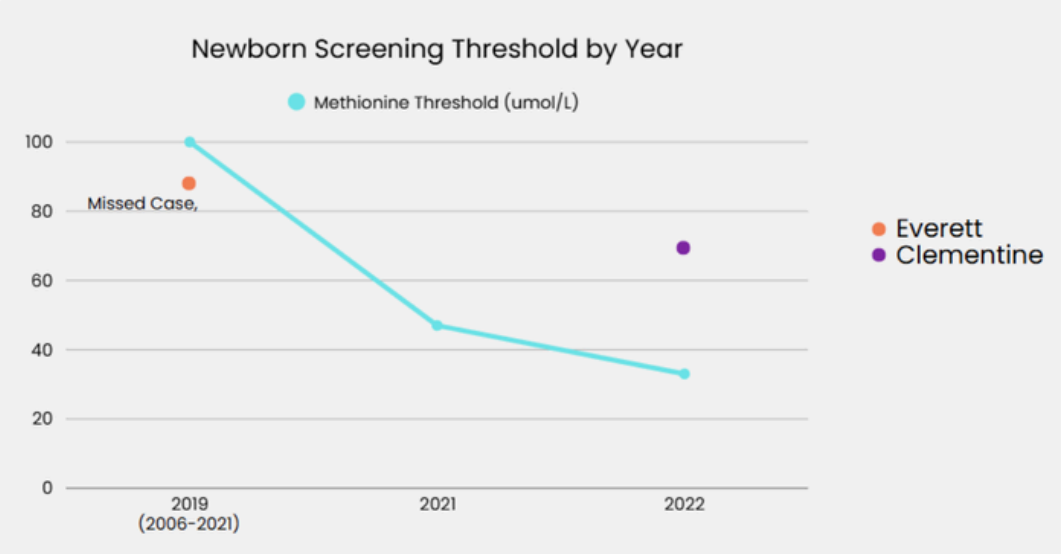
In fact, had Clementine not been born, we likely wouldn't have known Everett had HCU for years and we suspect the journey to obtaining a diagnosis would have been long and difficult. While we are so grateful for Clementine's early diagnosis, our story is a reminder that states can do better in their newborn screening for HCU. States can, and have, lowered newborn screen thresholds so that babies with HCU can be found! Thanks to Colorado lowering their threshold, Clementine and Everett were able to be diagnosed and treated for HCU.



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In terms of Everett's and Clementine's treatment for HCU, neither are b6 responsive. In addition to their formula, they both take betaine twice daily and methyl folate once daily. Both have been very responsive to the low-protein diet. We are happy to report that Everett's and Clementine's homocysteine levels are now in the 30s.

As for how Everett is doing, he is a happy, energetic, smart, and funny kid! He starts kindergarten in August at our local elementary school. He loves Transformers, Legos, and construction. He began his low-protein diet in April of this year and has been an absolute champ with changing his diet. He is learning how to recognize protein count on nutrition labels and understands he can only have up to a certain amount of protein each day.

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He drinks his formula drink twice a day, mostly without complaint (!), and says his new diet makes him feel a lot better. With respect to the impacts of his untreated HCU, we are still in the process of having him evaluated. He is quite tall for his age. He got glasses (for nearsightedness) shortly before he was diagnosed with HCU, and we have since learned he also has some lens dislocation due to his untreated HCU. Everett also had a lot of trouble regulating his emotions, experienced brain fog, and engaged in more solo play when his HCU was untreated. Now that he is being treated, he seems generally happier, more clear headed, and has been playing a lot more in big groups. His coordination, balance, and gross motor skills have significantly improved since receiving treatment for HCU, and he learned to swim and

ride a bike this summer. We are so proud of his progress!

As for Clementine, she is a smiley, happy 6-month-old. She loves rolling over and recently started sleeping through the night. Her favorite thing to do is watch her brothers play and you can tell she can't wait to be a big kid so she can join in the fun. She drinks a special HCU baby formula (made without methionine) mixed with regular baby formula – in specified quantities so that she gets just enough methionine to grow properly. She started eating solids a few weeks ago and is a big fan of all the fruit purees.

Both Clementine and Everett are doing very well! We know our journey with HCU has only just begun, but are grateful we were able to get their homocysteine levels within a manageable

range relatively quickly. We are hopeful there will one day be a cure and, until then, are doing the best we can under the circumstances and staying positive. We are lucky to work with the amazing Metabolic team at Children's Hospital Colorado, and to only be a short car ride away from the Metabolic Clinic. We are hopeful Clementine's and Everett's HCU diagnosis story will shed light on the importance of newborn screening and demonstrate that lowering the screening threshold can and will make a difference in the lives of children born with HCU!

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@hcunetwork
america.org

SEPTEMBER IS NEWBORN SCREENING AWARENESS MONTH!

What started as a PKU heel prick blood test developed by Dr. Robert Guthrie (whose own son had PKU!) in 1960, has evolved into the robust system that we know today.

Check out our 'Newborn Screening at a Glance' infographic to learn more!

Click here to download & share!

NEWBORN SCREENING at a glance

NEWBORN SCREENING IS A LIFE-SAVER

NBS is a public health program that screens newborns for **serious but treatable** health conditions.

Early detection and **timely treatment** can help prevent serious complications, such as developmental delays, illness, and death.

Approximately **12,000 babies** are diagnosed with a condition through newborn screening in the United States **each year.**

IT CONSISTS OF 3 PARTS

& IS PERFORMED WITHIN THE FIRST 24-48 HOURS OF LIFE

- blood test
- hearing screen
- heart screen

For the blood test, a healthcare provider pricks the baby's heel to collect **a few drops of blood**, which are then placed on a special paper and sent to a lab for testing. The blood test can detect certain genetic, metabolic, hormonal, and functional disorders

IT'S A SCREENING, NOT A DIAGNOSTIC TEST

If a screening test returns an out-of-range result, it indicates a higher risk of a particular condition, but **it does not confirm a diagnosis**. Further diagnostic testing is needed to confirm whether the baby actually has the condition.

NBS IS A FEDERAL PROGRAM, BUT VARIES FROM STATE TO STATE

These differences include the specific **conditions tested**, the **policies for follow-up**, and the **methods** used.

While most states screen for the majority of the RUSP* conditions, some states choose to screen for additional conditions not included in the RUSP. Conversely, some states may not screen for all RUSP conditions due to various factors like cost, infrastructure, or prevalence in the population.

RUSP = *Recommended Uniform Screening Panel; a list of conditions that the U.S. Department of Health and Human Services (HHS) recommends for inclusion in state newborn screening programs. The RUSP serves as a guideline for states to determine which conditions should be part of their newborn screening panels.*

Continued investment in our Newborn Screening programs ensures that **all babies** have **the best possible start in life.**

SEPTEMBER IS NEWBORN SCREENING AWARENESS MONTH!

Newborn Screening & Homocystinuria

50%

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

50

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.

The RUSP

(Recommended Uniform Screening Panel)

- The RUSP is a list of disorders that the Secretary of the Department of Health & Human Services *recommends* for states to screen as part of their state universal newborn screening programs.
- Disorders on the RUSP are chosen based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments.
- Each state ultimately determines what disorders its NBS program will screen for.
- Most states screen for the majority of disorders on the RUSP & some states also screen for additional disorders.
- Massachusetts was the first to adopt HCU newborn screening - in 1968! By 2009, all 50 states were screening for HCU.

https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp

Current Screening Method for classical HCU & its limitations

- Uses methionine level
- Methionine isn't elevated in most HCU patients in 24-48 hours after birth, especially those who are breastfed.
- Each state's newborn screening lab sets its own methionine cut-off to indicate a positive screen.
- A normal methionine level at birth is between 20-30.
- If a lab sets its methionine cut-off too high it will miss patients.

"Elliott's classical HCU was missed at newborn screening because his methionine level was not yet elevated enough to receive a positive screen. His level was 44 and to receive a positive screen in South Carolina, it must be 65."

-Liz Carter, mom of Elliott, diagnosed with classical HCU at age 2 1/2

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.

New developments give hope for the future

In 2023 the CDC came up with a 1st tier multi-plex approach that would allow total homocysteine to be the primary marker in screening for Homocystinuria.

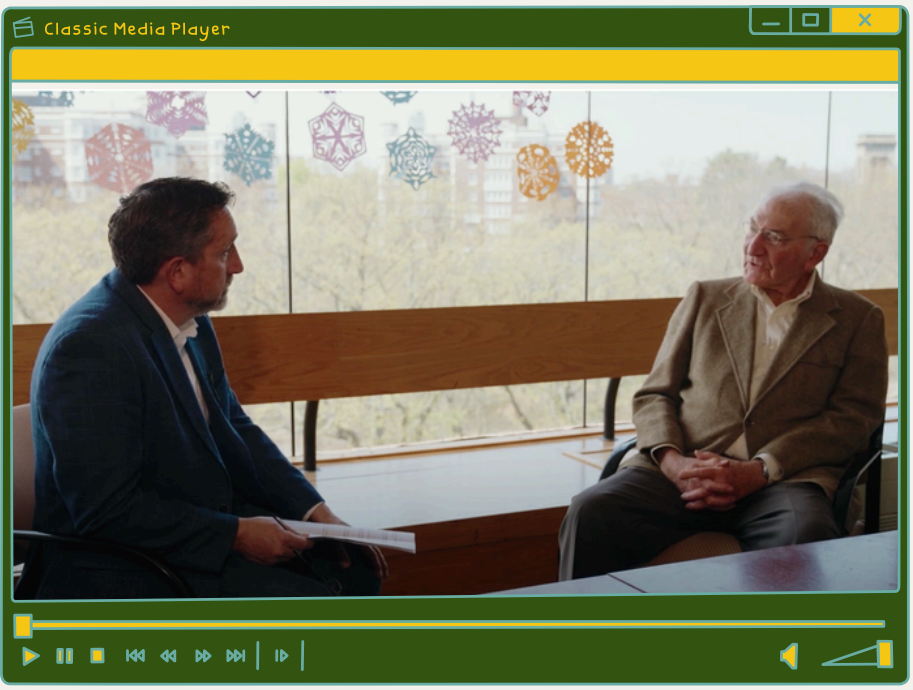
The goal? A more accurate screen, and identifying all babies with homocystinuria at birth.

Learn more:
https://hcunetworkamerica.org/newborn-screening/
https://academic.oup.com/clinchem/article/69/5/470/7068836?login=false

HCU NETWORK AMERICA

Click here to download & share!

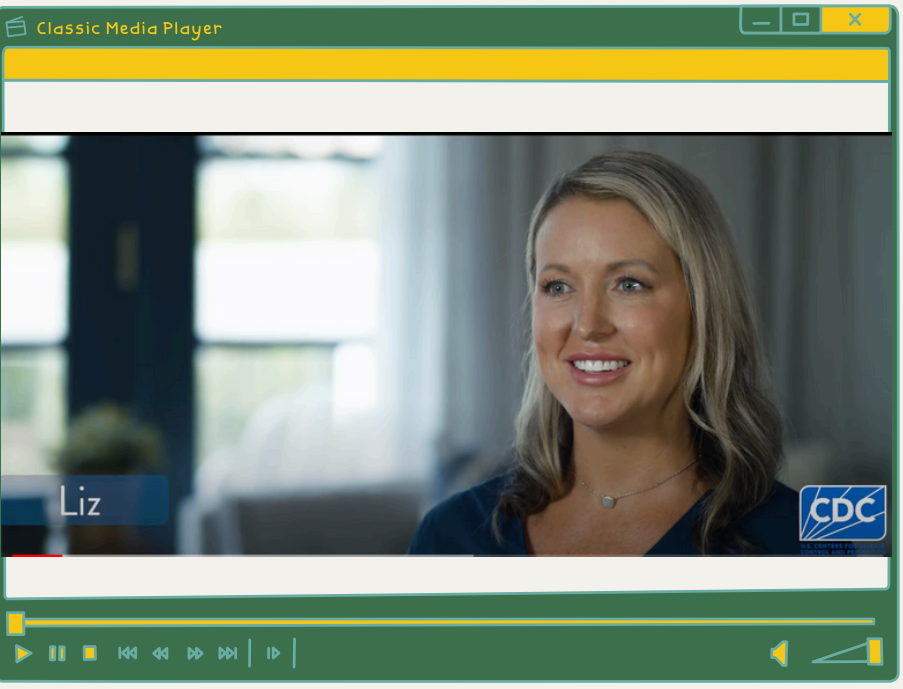
NEWBORN SCREENING AND HCU



History of Homocystinuria Newborn Screening with HCU Pioneer, Dr. Harvey Levy

HCU Network America Vice President Mark Lewis interviews the world-renowned Dr. Harvey Levy about his involvement and the history of newborn screening for the Homocystinurias.

Click to watch

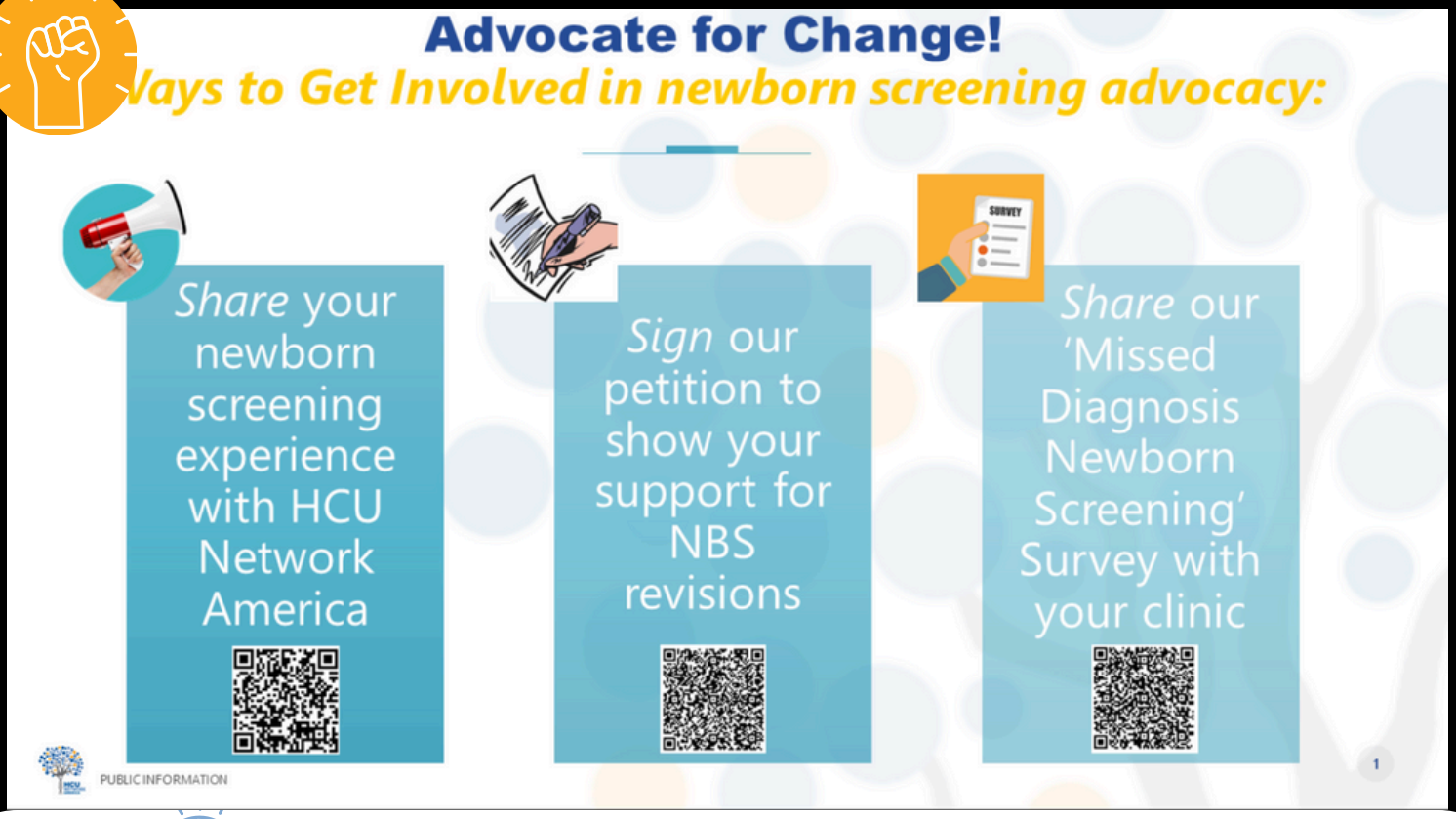


CDC Newborn Screening: “Homeward Bound”

Liz, the mom of a child with homocystinuria who was missed by Newborn Screening, shares her family’s story.

Click to watch

NEWBORN SCREENING



Advocacy Tips, Tricks & Ideas

- Share one (or both!) of our infographics, [along with something about your newborn screening story if you'd like], to social media during the month of September!
- Don't want to create your own posts for social media? That's ok! Keep an eye on HCU Network America's posts and share them!
- Check out Baby's First Test NBS Awareness Social Media Toolkit! It's got some ready-made posts and graphics that you can use!
- Use the hashtags: **#NewbornScreening** & **#2025NBS** to give your posts more visibility!
- Not a social media person? No worries! Make it a point to tell someone (*a friend, teacher, coworker, doctor, or even the barista at your coffee shop*!) that September is Newborn Screening Awareness Month and share a little bit with them about your (or your child's) newborn screening story.

UPCOMING EVENTS

UPCOMING EVENTS

HCU AWARENESS MONTH

VIRTUAL MEET UP

CLICK HERE!

SUNDAY 12 OCTOBER

4 PM CT/5 ET/ 3 MT

HCU AWARENESS MONTH IS COMING UP!

There are many ways that you can participate in HCU Awareness Month.

One way is by hosting a fundraiser!

Not only do fundraisers raise vital funds to help us support the Homocystinuria community, they spread awareness of the Homocystinurias and the challenges of living with the conditions.

Not sure how to get started?
Let our Fundraising committee help!

Email
info@HCUnetworkamerica.org

Click **here** for a list of additional HCU Awareness and Fundraising Event Ideas!

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

2025
NEWBORN SCREENING BOOTCAMP
September 17 I Virtual

Registration is now open for the 2025 Newborn Screening Bootcamp

Co-hosted by the EveryLife Foundation for Rare Diseases and Expecting Health, 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.

Register Here

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!

MEDICATION COMPARISON CHART

Medication Comparison

Understand Your Options

	NAME BRAND	GENERIC	COMPOUNDED	OVER-THE COUNTER
Example- Betaine	CYSTADANE® MANUFACTURED BY RECORDATI	BETAINE- MANUFACTURED BY ETON AND COSETTE	CUSTOM MADE TO DOCTORS ORDERS	PURCHASED THROUGH A 3RD PARTY RETAILER
Insurance Eligible	✓	✓	NOT OFTEN COVERED BUT DEPENDS ON INDIVIDUAL INSURANCE POLICIES. MANY PHARMACIES DO NOT ACCEPT INSURANCE	
Requires a Prescription	✓	✓	✓	
Cost ($=low to $$$$=high)	$$$$ COST IS HIGH DUE TO RESEARCH AND DEVELOPMENT OFTEN HAVE PATIENT ASSISTANCE PROGRAMS	$$ TEND TO COST LESS DUE TO RESEARCH AND DEVELOPMENT ALREADY BEING COMPLETED	$-$$$$ VARIED COST DEPENDING ON COVERAGE AND AVAILABILITY	$-$$$$ VARIED COST DEPENDING ON PRODUCT AND AVAILABILITY
FDA Approved	✓	✓	ACCREDITED THROUGH PCAB OR NABP	
Quality	HIGH QUALITY PRESCRIPTION GRADE	HIGH QUALITY PRESCRIPTION GRADE	MAY CONTAIN OTHER INGRIDIENTS VARIABLE QUALITY	MAY CONTAIN OTHER INGRIDIENTS VARIABLE QUALITY

HCU NETWORK AMERICA

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

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Das Spital der Eleonorenstiftung

Wands At The Ready...

Our Race for Research has officially kicked off!

It's still not too late to join the fun!

You can sign up (with as little as $5 to participate!) here!

Don't forget to log your miles!

Racers can sync their page to Strava OR visit your team's race page to log miles manually!

THANK YOU to our amazing sponsors for helping to make this year's Race for Research a success!

RECORDATI RARE DISEASES

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Learn more about Recordati Rare Diseases

RESEARCH FUNDED BY YOU!

Vasu Sethuraman, PhD

Investigating the Therapeutic Potential of Intestinal Methionine Elimination via Oral Enzyme Therapy in HCU

Kenneth Maclean, PhD

Combining taurine and formate/betaine treatment to improve clinical outcome and drug safety in classical CBS deficient Homocystinuria.

Thomas McCorvie, PhD

Crystallography-based fragment screening to develop pharmacological chaperones for classic homocystinuria

Ferdinand vonMeyenn

Establishing MTHFR- and Cobalamin G-deficient cerebral organoids: an in vitro disease model for investigating novel therapies

RESEARCH FUNDED BY YOU!

Dr. Brian Gilfix

Aminoglycosides for the Treatment of Inborn Errors of Vitamin B12 Metabolism

Devinder Kaur, PhD

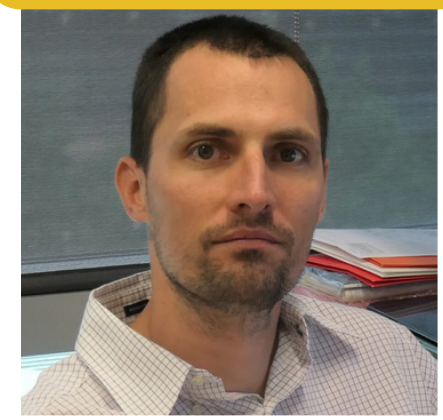
Development of Reference Ranges for Additional Newborn Screening Markers for Early Detection of the Homocystinurias: Classical Homocystinuria and Remethylation Disorders

Silvia Vilasi, PhD

Identification of Compounds to Rescue MMACHC Functional Deficiency in CBLC Disease

RESEARCH FUNDED BY YOU!

Tomas Majtan, PhD



Evaluation of benefits of thiol-based reductants in classical homocystinuria

Kenneth Maclean

New Metabolic strategies for improving the treatment of homocystinuria due to CBS deficiency and remethylation defects

Thomas McCorvie, PhD

Crystallography-based fragment screening to develop pharmacological chaperones for classic homocystinuria

MEET THE TEAMS!

 Amy's Army	Click here
Miles for Marley	Click here
 Miles for Andy	Click here
 Grayson's Gang	Click here
 Matteo's Muggles	Click here
Classical Champions	Click here
 Miles for Marcus	Click here
 Team Butter Stick	Click here
 Renna's Rare Runners	Click here
Team Recordati	Click here
 Carson's MTHFR Gene Team	Click here
CanPKU+ Racers	Click here

Recordati- Sylvia's Supporters	Click here
Team Will for HCU	Click here
Team Hawkins	Click here
 Team Anniston	Click here
 Team Hunt for Research	Click here
Brooke's Blazers	Click here
 Ellie's Entourage	Click here
Leo Frank	Click here
 Cure for Casey	Click here
ASA Walkers	Click here

Follow the leaderboard throughout the month!

https://charity.pledgeit.org/HCURaceforResearch2025

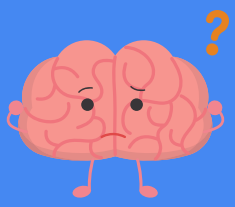
WHAT IS HOMOCYSTINURIA?

Homocystinuria denotes an elevation of homocysteine or homocystine in the urine.

Elevations in the blood may indicate an inherited form, but is not always the case.

Homocystinuria affects
1 in 200,00 people worldwide.

HOMOCYSTINURIA, THERE IS MORE THAN ONE?



B6 RESPONSIVE CLASSICAL HOMOCYSTINURIA

B6 NON-RESPONSIVE CLASSICAL HOMOCYSTINURIA

SEVERE MTHFR

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN C

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN D

HOMOCYSTINURIA, COBALAMIN D

HOMOCYSTINURIA, COBALAMIN E

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN F

HOMOCYSTINURIA, COBALAMIN G

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN J

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN K

METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA, COBALAMIN X

Classical homocystinuria is screened for in all 50 states, and most states screen for the various forms of methylmalonic acidemia with homocystinuria, however, it's estimated that at least 50 % of cases are missed! Without a diagnosis or early intervention, all homocystinurias can be fatal!

THERAPY?

The last approved therapy for the homocystinurias was approved in 1996.

Even with this therapy, patients remain at risk with high levels of homocysteine.

HELP ACCELERATE RESEARCH

10
Research Grants Funded

$418,000
Total research grants awarded and counting

MEET OUR NEW MEDICAL ADVISORS !

Dr. Andrew Lee

Assistant Professor at UC Irvine School of Medicine

Dr. Andrew Lee is an assistant professor in the department of neurology at UC Irvine School of Medicine. He completed his medical education at Boston University School of Medicine, his residency in neurology at New York University and fellowship in vascular neurology at Mount Sinai. Dr. Lee is board certified in both neurology and vascular neurology.

MEET OUR NEW MEDICAL ADVISORS !

Dr. Changrui Xiao

UCI Health neurologist

Dr. Changrui Xiao is a UCI Health neurologist with board certifications in neurology, medical genetics and genomics, and medical biochemical genetics. He specializes in caring for adults with neurogenetic and inherited metabolic disorders. His clinical interests include hereditary ataxia, neurometabolic disorders, inborn errors of metabolism in adults, genomic testing, and undiagnosed diseases. He is an associate editor of the journal Molecular Genetics & Genomic Medicine and a consulting editor for GeneReviews. His goal is to provide a medical home for adults with neurogenic and metabolic conditions, and to increase access to precision therapy for these conditions.
He earned his medical degree at Duke University School of Medicine in Durham, N.C. He completed an internship in internal medicine and a residency in neurology at the University of Chicago Department of Medicine in Chicago, Ill., where he also completed fellowship in clinical medical ethics and served as chief resident. He received additional training in medical genetics and medical biochemical genetics at the National institutes of Health and John's Hopkins University consortium program. He is currently the PI of UCI/CHOC Undiagnosed Disease Network Site".

MEET OUR NEW BOARD MEMBERS!

Kristen Skvorak

Dr. Skvorak is currently a freelance Preclinical Development consultant with 20 years of experience specializing in inborn errors of metabolism (IEM) and translational biology. She has dedicated her career to enabling the incorporation of the patient voice into drug development, from ideation through approval, and certifying the establishment of patient-centric strategies. She earned her PhD from the University of Pittsburgh in 2008, after which her research validating liver-directed cell therapies formed the foundation of two clinical trials for IEM in two countries (US and Sweden). She also contributed to the creation and characterization of the first viable model of Maple Syrup Urine Disease (MSUD), the intermediate MSUD mouse, and the first large animal model of Phenylketonuria (PKU), a porcine model. She also helped to further characterize a mouse model of PKU, illuminating new phenotypes not previously described. In 2016, Dr. Skvorak joined Codexis, an enzyme engineering company developing novel oral enzyme substitution therapies targeting metabolic disease and IEM. There, she contributed to the discovery and preclinical validation of several programs for oral delivery, including for PKU and Exocrine Pancreatic Insufficiency, which progressed to the clinic under Nestle Health Science. She also served as translational lead for preclinical programs in Homocystinuria and MSUD, which yielded peer-reviewed publications, multiple patents, and achieved Orphan Disease Designation and Rare Pediatric Disease Designation from the FDA, which are now moving forward with Syntis Bio. She has been consulting to support preclinical development of new therapies for IEMs and other metabolic diseases since 2023, serves on the Scientific Advisory Boards of two other IEAAM organizations, and is the Director of Research for the rare disease patient group CDG CARE.

MEET OUR NEW BOARD MEMBERS!

Brandon Tornes

CLICK HERE!

Brandon has lived most of his life in small towns in Wyoming and recently moved to Houston, Texas, with his wife Shandra, daughter Madyson, and son Mason. Six days after Mason was born, they found out through the newborn blood screening test that Mason had a metabolic disease, later confirmed to be Cobalamin C. Mason has the typical complications with Cobalamin C, including macular degeneration, but that doesn't stop him from living his best life! Mason is involved in the Miracle League (special needs baseball), Cub Scouts, and, most recently, Taekwondo!

Brandon enjoys playing his bass guitar, mountain biking, fly fishing, and being in the mountains. Living in small towns in Wyoming, he's learned to help those around him. Being a member of the Board of Directors allows him to share his story and show others going through a similar path in life that nothing can hold you back from what you want to accomplish.

You can read their family's full story here

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

Insert Ben welcomes Brittany to the table!

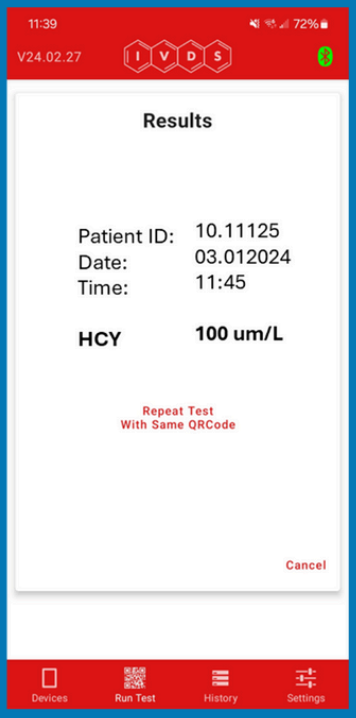
Brittany is the mother of Grayson, our August HCU Hero. She is also HCU Network America's Research Coordinator. Listen in as she shares not only about her role in this organization but also discusses her family's heroic journey with homocystinuria, Cobalamin G. Blurb here

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)”

CLICK HERE TO LEARN MORE ABOUT IVDS



IVDS

Harry Potter Butter"Beer"

Recipe Serves: 8 | Serving size: 1 Cup | Protein: 1 g

Ingredients:

- 2 liters of cream soda, chilled
- ¼ tsp caramel extract
- ¼ tsp butter extract
- 1 cup heavy whipping cream
- ¼ butterscotch topping
- ¼ powdered sugar

Equipment

- Mixer (stand, hand, or by-hand)
- Your most magical glasses
- Piping bags or zip blocks

Directions:

- In a large pitcher, mix cream soda, caramel, and butter extracts-- set aside
- In a bowl, use a mixer to whip the heavy cream until stiff peaks form
- Add butterscotch topping, powdered sugar, and mix
- Pour the soda mixture into your magical mugs
- Pipe butterscotch cream topping over the butterbeer
- Serve and enjoy!

Please do your research before consuming.

Low Protein Substitutions

For Heavy Whipping Cream

Nutrition Facts

About 32 servings per container
Serving size 1 Tbsp (15mL)

Amount per serving
Calories 45

	% Daily Value
Total Fat 5g	6%
Saturated Fat 3g	15%
Sodium 0mg	0%
Total Carbohydrate 0g	0%
Total Sugars 0g	
Includes 0g Added Sugars	0%
Protein 0g	

Not a significant source of trans fat, cholesterol, dietary fiber, vitamin D, calcium, iron and potassium.

CLICK HERE!

Please do your research before consuming.

HOMOCYSTINURIAS

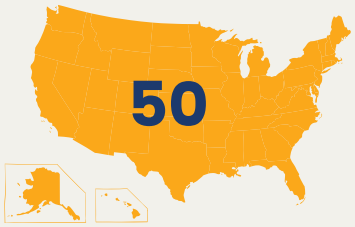
DATA COLLECTION PROGRAM

POWERED BY RARE X

Newborn Screening & the Homocystinurias

Approximate number of babies with Classical HCU that are missed at Newborn Screening

Some suggest up to 80% are missed!



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, 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.

CANPCU+ CANPKU+ HCU

HCU NETWORK AMERICA

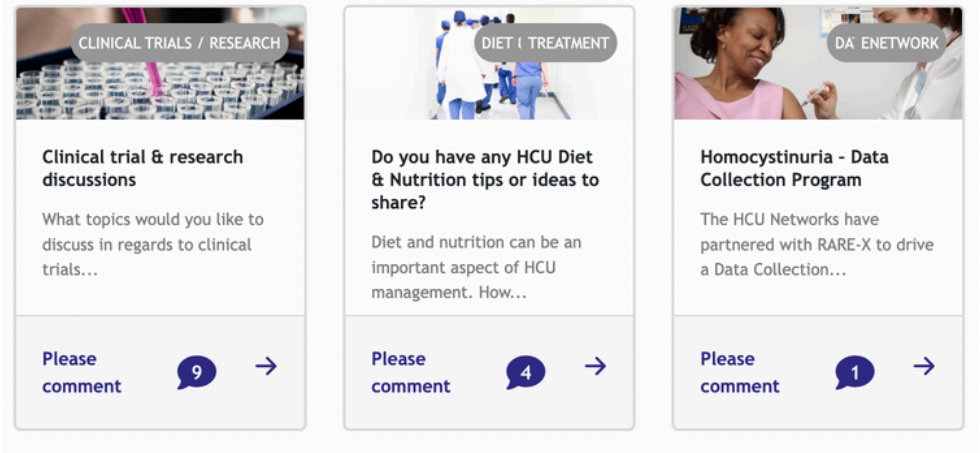
HCU Network Australia

JOIN THE HCU E-NETWORK

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.



HCU NETWORK AMERICA

HCU Network Australia

Join the conversation!

https://hcuenetwork.org/

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)

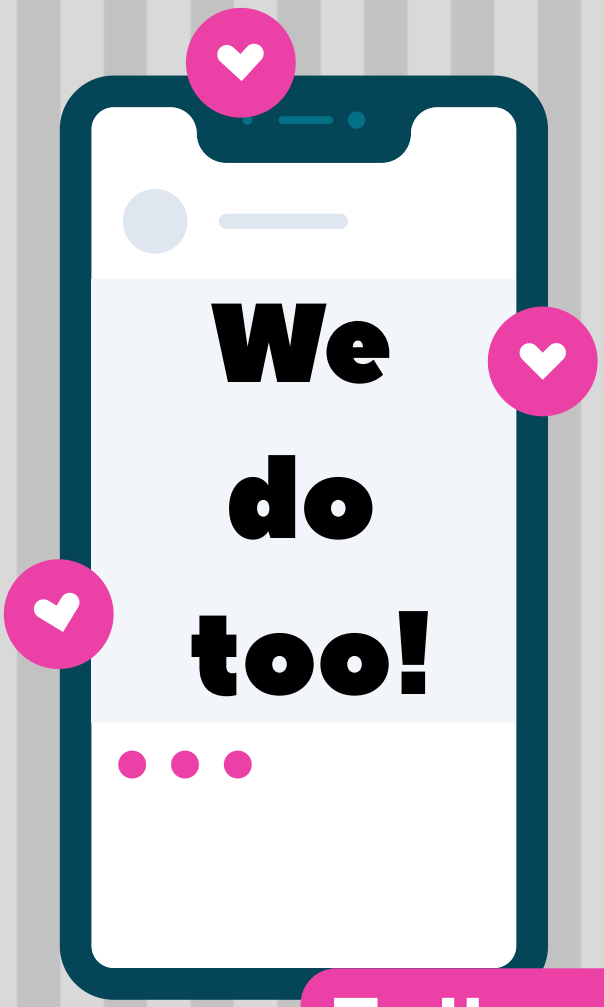
How?

- **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??



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