

The HCU Herald

October is HCU Awareness Month!

Featuring...

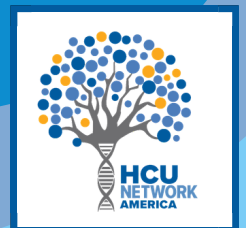


HCU Hero

**Kira from the
United Kingdom**



October 2026



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

HCU HERO: KIRA FROM THE UK



Kira was born in January 2017, slightly jaundiced, but otherwise a happy, healthy baby. She grew very quickly, going to the 90th centiles for height and weight within 6 months!

Life with Kira was generally the same as most other kids until she got an ear infection the summer of 2021, aged 4.5yrs. Although places were still locked down with Covid, Kira was still attending nursery (Dad was classed as an essential worker so it was permitted for her to go). She came home on a Thursday with a sore ear, feeling under the weather, then started being sick that night.

She seemed to perk up on the Saturday/Sunday but then went downhill again on the Monday. She couldn't keep anything down and was getting very lethargic. We spoke to our GP who recommended we bring her in the next day to be seen in person. Following some checks at the appointment on Tuesday the GP sent us straight to the CAT ward at the local hospital as Kira was dehydrated and her heart rate was a little low.

Me and my husband were both allowed to stay with Kira for a couple of hours while she was checked and admitted, but only one was allowed to stay the night with her due to Covid. I stayed with her that night and she seemed OK once she had an IV in, except she couldn't stop being sick every hour, even with anti-sickness medicines.

At around 10am on Wednesday, Kira had her 1st seizure in front of me and a student nurse who was changing some of her bandages on her IV. Kira was asleep at the time. The nurse went to get a doctor as my husband arrived, which is when Kira had her 2nd seizure whilst the doctor examined her. The doctor called for a crash team after noticing her pupils were mis-matched and unresponsive. What happened next is mostly a blur, but I've watched enough medical dramas to know what was happening to my baby girl. For a short time, we were huddled in the corner of the room watching various medical personnel attend to Kira and then were moved to a staff room to allow the doctors to work. We have never been so scared in our lives.

HCU HERO: KIRA FROM NEW YORK

It was very lucky we were already at the hospital. Within around 10-15 minutes (that felt like hours), Kira was conscious and talking, completely oblivious as to what had just happened. She was taken away for testing as they were naturally very concerned about what had happened to an otherwise healthy child. We sat in this suddenly very empty room, shell shocked, made some calls to family to let them know.

We were taken to ICU to see Kira a few hours later, she was being kept asleep following a CT scan. The doctors informed us that she had a very large blood clot in her brain that had caused a stroke. They needed to operate immediately to relieve the pressure and remove damaged sections of her brain. I signed some forms; we kissed her and then she went off to surgery. The staff put us up in a family room so we could stay at the hospital rather than having to travel back and forth.

We were warned that the location of the clot and damage may result in Kira being unable to talk properly and could affect movement in parts of her body. No-one would know until she woke from surgery and could be properly assessed.



At 5.45pm we got a call from her surgeon to say he had finished and everything looked a lot better than it had on the scans. It would be a couple more hours before she would be out and ready for us to visit. Unfortunately, we had fallen asleep when she came out of surgery (due to stress and fatigue) and woke to voicemails from the ICU staff telling us we could go see her. When we arrived, they were just bringing her round and removing the tube from her throat. Kira immediately asked for a drink of water- full sentence, very clearly. The staff were shocked at how clear she had spoken considering what had happened to her and so soon after surgery. She was also moving her hands and trying to push the staff away from her as she wanted to be left alone to sleep.

HCU HERO: KIRA FROM NEW YORK

The relief was profound! But we also didn't want to get our hopes up completely as we'd been told that recovery could be months to years. We were also informed at this point it wasn't one clot, but dozens of smaller ones that had become clumped together, but the hospital had no idea what caused it at the time.

As covid was still a factor at the time, my husband and I were not allowed to be in the hospital with Kira at the same time. We took it in turns spending 2-3 days with her, then swapping over so the other one could also spend 2-3 days with her.



Kira decided she was going to defy the odds (may have something to do with my husband promising to get her a big toy once she was well enough to leave the hospital). She was moving her arms and legs and talking within days (and to be fair may have done sooner but the amount of morphine she was on to begin with made her very drowsy). She was feeding herself within a week post op – with the smuggest grin on her face you have ever seen, especially around medical staff! At 7-10 days she was demanding we wheel her around the ward to see the staff as she was bored being stuck in the room watching Paw Patrol.

PT and OT had signed her off within 2 sessions as she could walk unaided and was beating them at giant connect four. She did get tired very quickly, which was natural and expected. At 2 weeks they were talking about sending us home for a short time as Kira was so healthy!

It was closer to 3 weeks when we took her home for the weekend – it felt like a dream! To be at home, the 3 of us together. We didn't do much other than rest, which we all needed, but it was amazing.

When we went back in after the weekend for some routine check-ups, we had another shock, we were told we'd be staying.

HCU HERO: KIRA FROM NEW YORK

As Kira had recovered so well, they wanted to do a 2nd surgery to put the piece of skull they had removed back where it belonged. Up until then it had been kept under her skin, near to her stomach. We were told this was the best place for it as it would keep the bone pieces 'alive' for a couple of months. So 2nd surgery, out of the blue. This went well but there were some complications in a day or two afterwards, so she did have to have a third surgery to correct these, but once that was done, she recovered very quickly again.

We got to the 4-week mark when they told us they had found a reason for it all- Classic Homocystinuria. We had never heard of it, even though it was tested at the newborn screening. (Kira tested negative at birth).

We left the hospital 4.5 weeks post op with a month's supply of Betaine, a prescription for Aspirin and Folic Acid, a referral for a wheelchair and a referral for the metabolic clinic.

Unfortunately for Kira, Betaine didn't work and she needed to go on to Anamix Junior and a low protein diet. Going from a high protein diet to 10 exchanges a day was a big shock for us all! Luckily Kira isn't a very picky eater and managed very well though the supplements were a harder thing to sort. She's amazing though and generally her levels are great with only the odd spike here and there (usually due to her growing).



We are very fortunate that we have amazing staff at the metabolic clinic who would happily answer our questions, offer advice, sort trials of prescription foods, and work with the GP to get them sorted. We also found the HCU and PKU communities online which also provided comfort – knowing there were others who were living with the condition helped.

Kira missed the start of Primary School as a result of this but was eager to start back and only missed about 4 weeks. The initial couple of weeks she had shorter days to ease her into it, but she fully settled in with ease and soon made friends and got on very well with everyone there.

HCU HERO: KIRA FROM NEW YORK

The staff have been amazing at helping us manage her diet and she had school meals with her classmates until year 3, with the school kitchen staff working with us directly to provide a variety of food that Kira could have. Kira has since decided she wanted packed lunches, like most of her friends.

We've had plenty of challenges along the way, but Kira won't be stopped by it. She does occasionally ask, 'why me', 'why can't I be the same as others' and there have been some tough questions to answer. We remind her, we are all different. Some of her friends can't eat dairy, some have asthma, some have diabetes. Plus, myself and my husband also have dietary intolerances too which Kira is aware of.

She is a bit unique though – when genetic testing was done, only one marker was found that she shares with me. Her Dad doesn't have any markers. Technically, Kira should not have HCU, but she does. The Doctors have said they will do new genetic tests at the 10-year mark to see what advancements in genetic testing find.

Kira knows the protein amounts of many different foods by heart, works out herself what she wants to use her exchanges on each day, and has a varied and healthy diet compared to most other kids we know. She loves Broccoli, Sausages, and Yorkshire Puddings.

Kira loves school and learning, always participating in the lessons, and helping other children. All the staff think she is amazing and so many kids and adults know her. She also attends an after-school club with kids of varying ages from schools around the area and is very well known to them. She loves helping to look after the much younger kids there. She's a happy, healthy, (very tall) 9.5yr old who loves K-Pop Demon Hunters! This year a poem she wrote was published in a book with other children from around the country (the book can be found in the British Library). We are beyond proud of Kira. She has met every challenge head on and keeps pushing herself, testing her limits and surpassing them with determination.



October is HCU AWARENESS MONTH!



Week 1: Intro to the Homocystinurias

Week 2: Focus on Classical HCU

Week 3: Focus on Cobalamin Defects

Week 4: Focus on Severe MTHFR

Week 5: Recap

#Homocystinuria

#HCUAwareness

#HOPECONNECTSUS

#GOBLUEFORHCU

How can you spread awareness?

MONDAY



Re-share
our infographic
for the week.

TUESDAY



Share the
incidence rate
of your
condition.

WEDNESDAY



Create an
image with
you and
arrows pointing
to your
symptoms.

THURSDAY



Make a video
or share a photo
of what your
treatment
looks like.

FRIDAY



Share your
patient story.

HCU AWARENESS MONTH
OCTOBER



HCU
NETWORK
AMERICA

HCU Awareness Month

Activity List

Small
Actions.
Big Impact. 

ENGAGE / EDUCATE / MAKE AN IMPACT

- | | |
|---|--|
| ☐  Change your social media picture to the HCU Awareness Ribbon | ☐  Real cost of HCU: Grocery Cost Comparison #Medical Nutrition Equity Act, or share your patients with HCU require injectable B12. B12 on average is \$300-400 a month and most insurance companies don't cover it! |
| ☐  Start a HCU fundraiser | ☐  Share a picture or video capturing all the medications you take (this includes formula for those who need it). |
| ☐  Share an infographic about HCU | ☐  Share a picture of your first pair of glasses, or a device that helps you navigate or communicate due to lack of vision |
| ☐  Share a patient story | ☐  Share something you wish people understood about HCU |
| ☐  Share your diagnosis story | ☐  Wear jeans for your rare genes #ItsInOurGenes |
| ☐  Challenge your friends to the same amount of protein and three normal protein shakes a day #ToastToatHCU | ☐  Wear your HCU Shirt and share a pic online – #HopeConnectsUs |
| ☐  Share a pic of an item that has the same amount of protein you can have | ☐  #GoBlueforHCU |
| ☐  Share your daily diet record – completed | ☐  #HCUAwareness post in a public place |
| ☐  Share a low-protein meme | ☐  Share with a stranger what HCU is and why it's important to you |
| ☐  Share your favorite low-protein recipe! Bonus if you cook it and share a pic | ☐  #Create4Cure – Create a work of art that brings awareness for HCU – can be a song, dance, a painting – get creative! |
| ☐  Dining out, low protein style. Where do you like to eat? | ☐  #High5forHCU – List 5 ways HCU makes you a stronger, better person |
| ☐  Share a pic of what your grocery store haul looks like | ☐  All states test for classical HCU. But many are still missed |
| | ☐  Share a picture of you and a HCU buddy! Or tag a friend who is of great support |
| | ☐  #FacesofHCU – Share a picture of you saying, "I am one of the 1 in 200,000 people with HCU" |
| | ☐  #Hope4HCU – Share 4 things that give you hope and encouragement |
| | ☐  Share the HCU timeline – if you know other facts, let us know! |
| | ☐  Cutting Edge of HCU: Share about a therapy that is in the works! |

Together we can
make a difference! 

To find additional information and resources, visit:
<https://hcunetworkamerica.org/hcu-awareness-month/>



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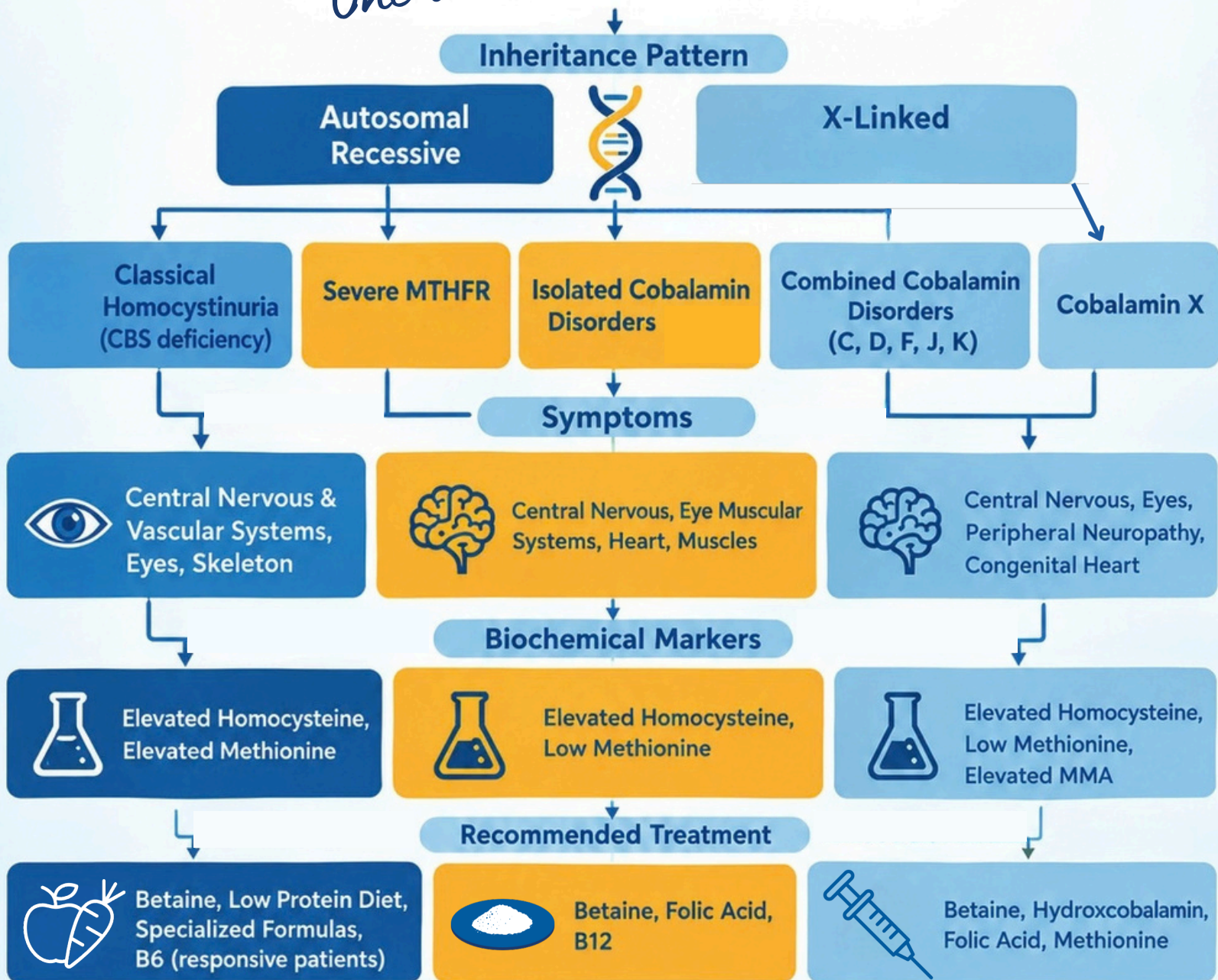
HCU AWARENESS MONTH

| OCTOBER



Homocystinuria

One biomarker, many conditions



HCU AWARENESS MONTH
OCTOBER





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How rare is **HCU?**

1 in 200,000

PEOPLE IN AMERICA
LIVING WITH HCU.

That may be rare...
but for the people
living with it,
HCU is real life.



HCU AWARENESS MONTH | OCTOBER

— RARE DISEASE. REAL PEOPLE. REAL IMPACT. —

HCU isn't the same as **HYPERHOMOCYSTEINEMIA**

They sound similar, but they are different conditions with different causes and impacts.



HOMOCYSTEINURIA (HCU)



Genetic disorder

Caused by a change in specific genes.



Homocysteine builds up

The body can't properly process it.



Can cause multisystem complications

Affects the eyes, brain, heart, blood vessels, bones and more.

VS

HYPERHOMOCYSTEINEMIA



Elevated homocysteine

Higher than normal levels in the blood.



Can have many different causes

Including nutritional deficiencies, other medical conditions and medications.



Not the same as HCU

Does not include the genetic disorder or the full range of complications.

Know the difference.

EARLIER RECOGNITION MATTERS.

HCU AWARENESS MONTH

| OCTOBER





A SIMPLE
AMINO ACID SCREEN
OF BLOOD AND URINE CAN
HELP INDICATE HCU



HCU AWARENESS MONTH
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— RARE DISEASE. REAL PEOPLE. —
REAL IMPACT.

HCU can affect MULTIPLE SYSTEMS of the body.

*One biomarker,
many conditions.*

HCU can impact many parts
of the body, including:



Brain & Nervous System

Developmental delay,
stroke, seizures, mental
health



Eyes

Vision loss,
maculopathy,
nystagmus



Heart

Blood clots,
enlarged aorta,
cardiac complications



Liver

Liver damage



Muscle

Weakness,
low muscle tone



Blood & Clotting

Blood clots,
thrombosis



Lungs

Blood clots,
pulmonary issues



Digestive System

Feeding challenges,
nutritional deficiencies



Kidneys

Kidney damage,
TMA, HUS



Skeletal System

Long limbs,
tall stature, curved spine,
low bone density



Skin

Dermatitis, rash

HCU AWARENESS MONTH | OCTOBER



**HCU
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AMERICA**

THE JOURNEY OF Homocystinuria

FROM DISCOVERY TO A BRIGHTER TOMORROW

Early detection.
Better treatment.
Brighter futures. ♥

1960s - 1999

HCU - WE'VE COME A LONG WAY...



1962

- Dr. Gerritsen, Carson and Neill describe Homocystinuria as a cause of intellectual disability by urine amino acid screen.
- Dr. George Spaeth independently diagnoses a patient with Classical Homocystinuria.



1964

- Dr. Harvey Mudd describes that cystathionine synthase enzymatic deficiency causes homocystinuria in Science.
- Guthrie test can detect high methionine for NBS.
- Low Met diet known to reduce levels and symptoms.
- Doctors prescribe blend of amino acids with low Met diet.



1968

- Folic acid found to help manage Classical HCU.



1969

- Dr. Barber and Spaeth report pyridoxine (vitamin B6) responsive patients.



1969 & 1970

- Dr. Mudd and Harvey Levy describe the enzymatic defect causing cobalamin C deficiency.



1970

- Dr. Goodman describes response to B12 therapy in Cobalamin C patients.



1981

- Betaine found to help reduce hcy levels.



1983

- B6, B12 and folic acid found to better treat high hcy levels.



1988

- Dr. Muenke and Kraus map the CBS gene.



1991

- Organic Acidemia Association receives non-profit status.



1993

- Dr. Kraus describes the human cystathionine beta-synthase cDNA.



1995

- Dr. Watanabe describes the first CBS knockout mouse model.
- In 2004 Dr. Kruger develops different transgenic mouse models to improve their survival.



1996

- Cystadane (betaine anhydrous) obtains FDA approval for CBS deficiency, MTHFR deficiency and cbl defects.

2000 - Present

HCU - WE'VE COME A LONG WAY...



2006

- Dr. Lerner-Ellis and Rosenblatt et al identify the gene for methylmalonic aciduria and homocystinuria, cblC type.
- High dose hydroxocobalamin therapy shows benefit in cblC patients.



2014

- First mouse model of cblC is described, but is lethal in embryonic life.
- HCU Network Australia founded.



2015

- Different cblC mouse models are developed using gene editing that survive pregnancy and delivery, but die within first 30 days unless rescued with hydroxocobalamin or gene therapy.



2016

- HCU Network America founded.
- Dr. Kraus's lab reports enzyme replacement with PEGylated CBS shows promising results in a murine model.
- Dr. Manoli et al show methionine restricted medical foods and protein restriction can negatively affect brain growth in cblC.



2017

- Dr. Morris et al describe guidelines for CBS deficiency.
- Dr. Huemer et al summarize guidelines for cobalamin-related remethylation disorders.



2018

- HCU Network America host first patient/expert conference.
- Human trials for enzyme replacement therapy start, research for other new therapies progresses.



2019

- Intensification of hydroxocobalamin therapy in cobalamin defects in the newborn period shows positive result.

But we need to keep working
until a cure is found for all ♥



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OCTOBER



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AMERICA**

*Together we can
make a difference!* 

10% OFF
Go Blue For HCU Shirts

**IN THE HCU NETWORK AMERICA
BONFIRE STORE**



<https://www.bonfire.com/store/hcu-haberdashery/>



Use Code

ANNIVERSARY10

through
October 31, 2026



*Wear your support.
Spread awareness.
Make an impact.* 



SHOP NOW 

Support HCU Awareness.
Support the Community.

**HCU
AWARENESS
MONTH**
OCTOBER

Matching Gift Challenge

In honor of our 10-year anniversary, the **Lewis Family, Margie McGlynn**, the **Bonucci Family**, and the **Kelly Family** have come together to offer a **\$40,000 match**.



During the winter holidays, warmth and generosity can be felt near and far. We ask every patient and family to help us raise funds to continue our homocystinuria community outreach, education, and advocacy initiatives to educate providers and be there for families like Everett and Clementine's. Please share our appeal letter with your colleagues, friends, and family.

Three Easy Ways to Donate

[Click here to donate](https://bit.ly/HCUMatch2026)

<https://bit.ly/HCUMatch2026>



By check
HCU Network America
15 S. Mallory Ave
Batavia, IL 60510

EMPLOYER MATCHING GIFT PROGRAM

Did you submit for your Employers Corporate Matching Gifts Program?

What are Matching Gifts?

Corporate matching gifts are a type of gift giving in which companies financially match donations that their employees make to nonprofit organizations. When an employee makes a donation, they need to request the matching gift from the employer, who then makes their own donation. Companies usually match at a 1:1 ratio, but some will match 1:2 or 2:1.

Why are Corporate Matching Gifts Valuable?

Corporate matching gifts are valuable because they are free money for your nonprofit of choice, HCU Network America! Your donation has double the power without you having to give double the amount.

Does HCU Network America Really Benefit?

If corporate matching gifts weren't valuable, we wouldn't be sending this letter. In 2020 we received approximately \$45,000 in corporate matching gifts!

How Do I Find out if my Employer has a Corporate Matching Gifts Program?

Your company website or websites such as doublethedonation.com make it easy for you to search and see if your employer takes part in corporate matching gifts. If you can't find it online, please consult with your employee handbook, HR rep or manager to find out.

How Do I Request my Donation is Matched by my Employer?

1. The donor completes their donation
2. The donor submits matching gift request
3. Company reviews donation and nonprofit eligibility and reaches out to nonprofit
4. Nonprofit verifies the donation was made
5. If eligible, the nonprofit will receive the matching gifts request!

Top Matching Gift Companies

Company Match Ratio

- | | |
|-------------------------|------------------|
| • General Electric 1:1 | • Microsoft 1:1 |
| • Gap Corporation 1:1 | • Pfizer 1:1 |
| • ExxonMobil 3:1 | • Coca-Cola 2:1 |
| • Johnson & Johnson 2:1 | • And many more! |

Did you know some companies match retired employees donations?

PREGNANCY

PLANNING GUIDE

for Women with Homocystinuria

Many women with well-managed homocystinuria have successful pregnancies and healthy children. The key is planning **ahead**, **optimizing metabolic control** before conception, and receiving **coordinated** care from both a metabolic specialist and a maternal-fetal medicine team throughout **pregnancy** and the postpartum period.

1

BEFORE BECOMING PREGNANT

- Optimize metabolic control—keep total homocysteine as well controlled as possible
- Maintain stable dietary management
- Stay consistent with medications
- Review all treatment with your care team
- Determine who you need on your care team; maternal fetal medicine physician
- Begin prenatal vitamins if advised
- Gather a support network

QUESTIONS TO ASK YOUR CARE TEAM

- Are my current homocysteine and methionine levels in a safe treatment zone?
- If not, what adjustments need to be made?
- Should any of my medications be adjusted?
- When do I need to be seen by maternal fetal care? (for high risk pregnancy)
- Will I need anticoagulation during pregnancy or after delivery?
- How often will labs be monitored?
- How should I adjust my diet and formula?
- What adjustment do I need to make if I choose to breastfeed?

2

DURING PREGNANCY

- Increased risk of blood clots
- Talk to your doctor about anticoagulants
- Consider your individual history and risk factors
- Expect more frequent lab monitoring
- Nutritional needs change—protein and methionine tolerance may change during pregnancy

3

NEWBORN PERIOD

6-8 WEEKS POST-PARTUM

- Blood clot risk remains elevated
- Sleep disruption and recovery can affect adherence
- Nutritional needs continue to change
- Maintain medication protocol and diet



KEY CARE CONSIDERATIONS

Partnering with your care team before, during, and after pregnancy helps support your health and the best possible outcomes for you and your baby.

**As soon as you find out you are pregnant,
immediately contact your care team**



You are not alone.

With planning, support, and expert care, you can feel confident on your pregnancy journey.



HOMOCYSTEINURIA & Pregnancy

Quick Reference for Obstetricians, Family Physicians & Maternal-Fetal Medicine Providers

What Is Classical Homocystinuria?

A rare inherited disorder caused by cystathionine β -synthase (CBS) deficiency, resulting in markedly elevated total homocysteine (tHcy) and increased risk of:

- Venous and arterial thrombosis
- Stroke
- Pulmonary embolism
- Lens dislocation
- Osteoporosis
- Neurocognitive complications

Pregnancy is high-risk because HCU and pregnancy both increase thrombotic risk.

Before Conception

Goals:

- Optimize metabolic control (tHcy <100umol/L)
- Ensure adherence to medication, diet and/or formula
- Review medications and supplements
- Obtain baseline:
 - tHcy
 - Methionine
 - Nutritional status
 - B12/Folate
- Early referral to Maternal-Fetal Medicine if available
- Open communication with metabolic specialist and metabolic dietician

During Pregnancy

Major Concern: THROMBOSIS

Patients with HCU have an increased lifetime risk of thrombosis. Pregnancy adds additional hypercoagulable risk.

Consider:

- Early hematology consultation
- Individualized thromboprophylaxis plan
- Close communication with metabolic specialist and metabolic dietician
- Monitor
 - Total homocysteine (tHcy)
 - Methionine
 - Maternal nutrition
 - Fetal growth

Postpartum Period

Highest-Risk Time for Clots

The first weeks after delivery may carry significant thrombotic risk.

Key actions:

- Maintain metabolic therapy
- Continue recommended thrombosis prevention plan
- Ensure close postpartum follow-up
- Close communication with metabolic specialist and metabolic dietician

A woman with well-controlled HCU can have a successful pregnancy.

Call the Metabolic Team If:

- tHcy rises substantially
- Unexpected dietary or medical formula changes
- Hyperemesis prevents intake of medications or medical formula
- New or worsening neurologic symptoms develop
- Suspected thrombosis occurs
- Delivery planning questions arise



Treatment Guidelines

How Accurate Are Your Homocysteine Labs?

Does Your Lab Follow Protocol?

Recommended Collection Protocol

Immediately place on wet ice
Centrifuge specimen within 4 hours
If not immediately placed on ice, plasma must be removed from cells within 1 hour
Refrigerated centrifuge is not required

Why is it important?

Homocysteine continues to be released from red blood cells after collection, **causing falsely elevated results**

Levels can increase 10% per hour at room temperature if not processed

Important Considerations



Timing and Consistency! Try to have labs drawn at roughly the same time of day, under similar conditions and according to your metabolic clinic instructions.



Trends Matter! One elevated value is not always an emergency. Consider looking at several months of results.



You may see it reported as: tHcy, total plasma homocysteine, or Homocysteine total

- A total homocysteine (tHcy) test is a blood test that measures the total amount of homocysteine in the blood, including both free homocysteine and homocysteine bound to proteins.
- Homocystine is not a useful laboratory value for the management of HCU.

Total Homocysteine vs Homocystine

Total Homocysteine

Measures ALL Homocysteine present in blood
Normal values=5–15 umol/L

Homocystine

Measures only specific oxydized form
Normal values= <1 umol/L
Typically in amino acid profile (US)

- Keep track of your own levels to identify patterns over time
- Knowing your personal goal is often more useful than simply knowing whether a result is "high" or "low"
- Discuss your lab results with your doctor before making any changes





CARETAKERS GUIDE TO HCU



? What is HCU?

Homocystinuria (Ho-mo-cys-tin-uria), or HCU, is a rare inherited metabolic condition. People with HCU cannot break down the amino acids **methionine** (me-thahy-uh-noon) and **homocysteine** (hō-mō-'si-stā-ēn) in their bodies.

Methionine (Met) is found in most foods that contain protein. HCU is a severe medical condition that can be treated with a special HCU medical formula, a diet low in protein and Met, and some vitamins and other medicines.



Cause and Effect

Normally Methionine breaks down into another amino acid, homocysteine (HCY) (hō-mō-'si-stā-ēn). The byproduct homocysteine (HCY) also builds up and has very unhealthy and dangerous side effects when protein (more specifically Met) is ingested. High HCY levels are harmful to the eyes, skeletal, vascular and central nervous systems.



To help prevent these issues, those with HCU must follow a special diet with low protein and drink their HCU formula throughout the day.

Possible Complications of High HCY

- Severe nearsightedness
- Lens dislocation
- Cognitive deficits
- Behavioral problems
- Clumsiness
- Long limbs
- Blood clots
- Strokes



What is a low protein diet?

The majority of the HCU diet is composed of fruits, vegetables and specialty low protein foods.

May still need to be counted

- Fruit: Apples, Papaya, Pears, Strawberries, Tangerines
- Veggies: Bell Peppers, Cabbage, Celery, Eggplant, Tomatoes
- Butter and Oils
- Medical Low Protein Food



Allowed, but in limited quantities

- Fruit: Avocado, Figs, Jackfruit, Kiwi, Oranges
- Veggies: Brussel Sprouts, Corn, Peas, Potatoes
- (Some) Sugary Cereal
- Popcorn



Not allowed unless okayed by parent

- Meat
- Nuts
- Grains
- Most Dairy
- Beans
- Eggs
- Legumes



This information is not intended to take the place of medical advice or care you receive from your health care professional and intended for information purposes only. Permitted foods and quantities will vary. For a full list of permitted foods, please consult the child's metabolic care team.



To learn more about Homocystinuria, please visit: <https://hcunetworkamerica.org>

Support

- ♥ Treat them as any other child
- ♥ Teach them following a diet is important
- ♥ Encourage them to drink their formula
 - ♥ Don't make comments about the smell or flavor
- ♥ Don't deviate from the allowed foods, even if it's "just a bite"
- ♥ Attend homocystinuria events



Communicate

-  Communicate with parents and ask questions
-  Communicate what they have eaten, have not eaten, or if you plan to feed them anything - ask in advance. Make sure the items you have selected fit their protein allowance.



Remember

- ★ They are not sick
- ★ It's not an allergy. You will not see immediate side effects if the diet isn't followed
- ★ Damage from not following the diet tends to be irreversible
- ★ It's a diet for life!



Learn

-  Learn to properly read food labels
-  What foods they can and cannot eat and their exact protein and methionine contents
-  Learn how to weigh and measure foods
-  Learn how to cook low protein foods
-  Learn how to make their formula
-  Learn more about Homocystinuria





First Revision of the Guidelines for the Diagnosis and Management of Remethylation Disorders

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Why were the guidelines updated?

The original international guidelines were published several years ago. Since then:

- More patients have been identified through newborn screening.
- Doctors have gained experience treating patients earlier and for longer periods.
- New evidence has become available about medication dosing, monitoring, and long-term outcomes.
- Patient organizations participated in the guideline development process, helping ensure that patient and family perspectives were included.

The 2026 revision reviewed the latest available evidence and expert experience to provide updated recommendations.

KEY UPDATES

1. Stronger emphasis on early diagnosis

The revised guidelines highlight that early diagnosis, especially through newborn screening, is associated with better outcomes. Children identified before symptoms develop generally have a better chance of avoiding serious complications.

2. More detailed recommendations for laboratory testing

The updated guidelines stress measuring:

- Total homocysteine
- Methionine
- Methylmalonic acid (MMA), when appropriate
- Vitamin B12
- Folate levels

These tests help doctors distinguish remethylation disorders from other conditions and monitor treatment effectiveness.

3. Greater support for higher-dose hydroxocobalamin treatment

One of the most important updates is growing evidence that earlier use of higher-dose injectable hydroxocobalamin (a form of vitamin B12) may improve:

- Brain development and cognitive outcomes
- Neurological symptoms
- Eye disease in severe cobalamin-related disorders such as cblC
- The guideline notes emerging evidence supporting doses above 0.35 mg/kg/day in some patients

4. Continued recommendation for betaine therapy

For severe MTHFR deficiency and many other remethylation disorders, betaine remains a cornerstone treatment because it helps lower homocysteine and supports methionine production.

5. Clearer treatment goals

The revised guidelines recommend trying to keep:

- Homocysteine levels as close to normal as possible
- Methionine levels in a healthy range
- MMA levels controlled when relevant

These laboratory targets are increasingly recognized as important markers of good metabolic control.



What Does This Mean for Families?

There is reason for optimism

The revised guidelines reflect significant progress since the original recommendations. Experts now have more evidence that:

- Early diagnosis improves outcomes.
- Treatment can reduce illness and death.
- Earlier and potentially higher-dose hydroxocobalamin may provide additional benefits.
- Long-term management is helping more patients reach adulthood and achieve improved quality of life.

The importance of lifelong follow-up remains unchanged

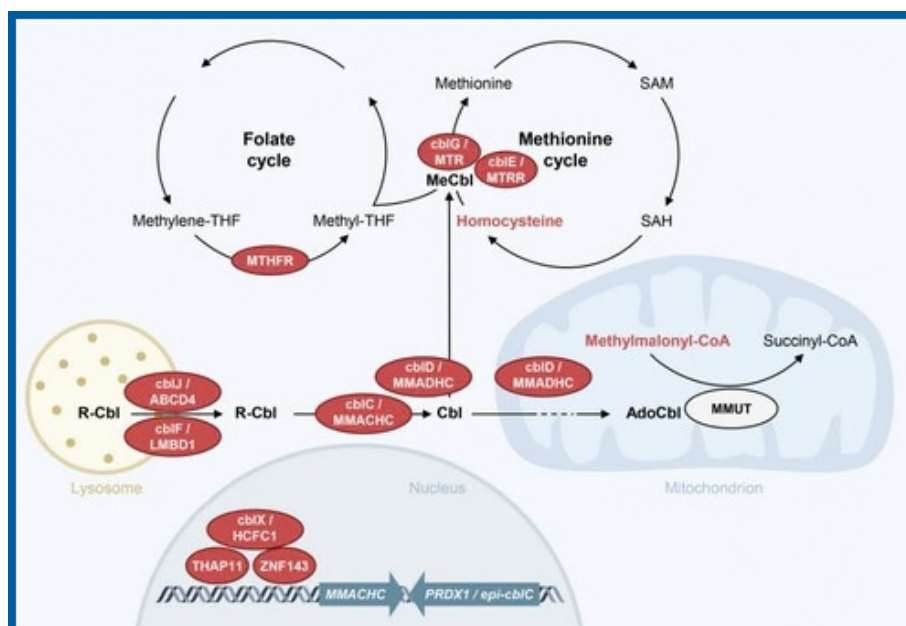
Even when a patient appears stable, ongoing follow-up is essential because these disorders can affect multiple organs over time. Monitoring vision, development, neurological function, kidney health, and laboratory values remains important.

Access to treatment remains a challenge

The guideline specifically notes that obtaining highly concentrated hydroxocobalamin formulations can be difficult in many regions. This remains an important unmet need for patients and families.

The 2026 guideline update is encouraging news for families affected by cblC, MTHFR deficiency, and other remethylation disorders. The biggest messages are:

- Diagnose patients as early as possible, ideally through newborn screening.
- Start effective treatment promptly.
- Continue using betaine and injectable hydroxocobalamin as core therapies.
- Aim for better biochemical control of homocysteine, methionine, and MMA.
- Consider emerging evidence that higher-dose hydroxocobalamin may improve neurological and eye outcomes, particularly in severe cobalamin-related disorders.
- Maintain lifelong monitoring with an experienced metabolic team.
- Overall, the revised guidelines show that earlier recognition and more aggressive treatment are helping improve outcomes for many individuals with remethylation disorders



We Went To *SSIEM!*



HCU Network America was proud to attend SSIEM 2026 in Helsinki, Finland, connecting with researchers, clinicians, advocates, and members of the global metabolic disease community. The conference provided an opportunity to highlight important research in homocystinuria, share the patient perspective, and strengthen connections that help move HCU research forward.

Final Scores



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Team Name	\$	Miles
Hunt for Research	\$28,249	160.6
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Classical Champions	\$1,915	620.17
Amy's Army	\$3,175	0
Team Cobalamin C	\$1,715	26.1
Team Butterstick	\$1,345	1.1
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Team Hawkins	\$1,268	268
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CanPKU+ Racers	\$530	61
Never Lewis Hope	\$2,465	100.7

Final Scores



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Axelotyl	\$600	5.5

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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!



The latest episode



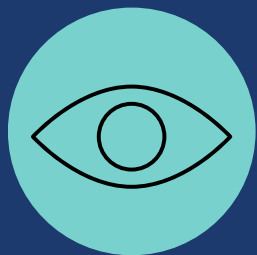
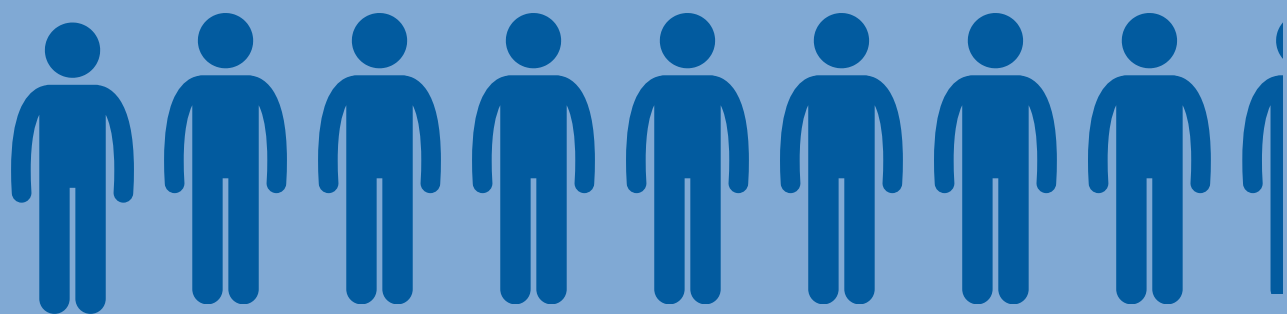
In this episode, Ben talks with Joseph, whose son Matthew faced developmental delays and was eventually diagnosed with homocystinuria (HCU) and neurofibromatosis type 1 (NF1). Joseph discusses the challenges of a delayed diagnosis and how their experience motivated the family to advocate for rare disease awareness. This conversation highlights the significance of newborn screening, trusting parental instincts, and assisting families in finding answers more quickly.

HOMOCYSTINURIAS

DATA COLLECTION PROGRAM

POWERED BY RAREX

Of the 81 participants who took the Head-to-Toe Survey
Here are the top 5 areas the data is pointing too



53
Vision Issues



48
Brain/Nervous
System



36
Bones/Cartilage
/Connective
Tissue



36
Sleep



40
Digestive
Issues

Complete the Head to Toe Survey at
<https://homocystinuria.rare-x.org>



HCU
NETWORK
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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!*

- **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?

Why?

Contact Register

How?

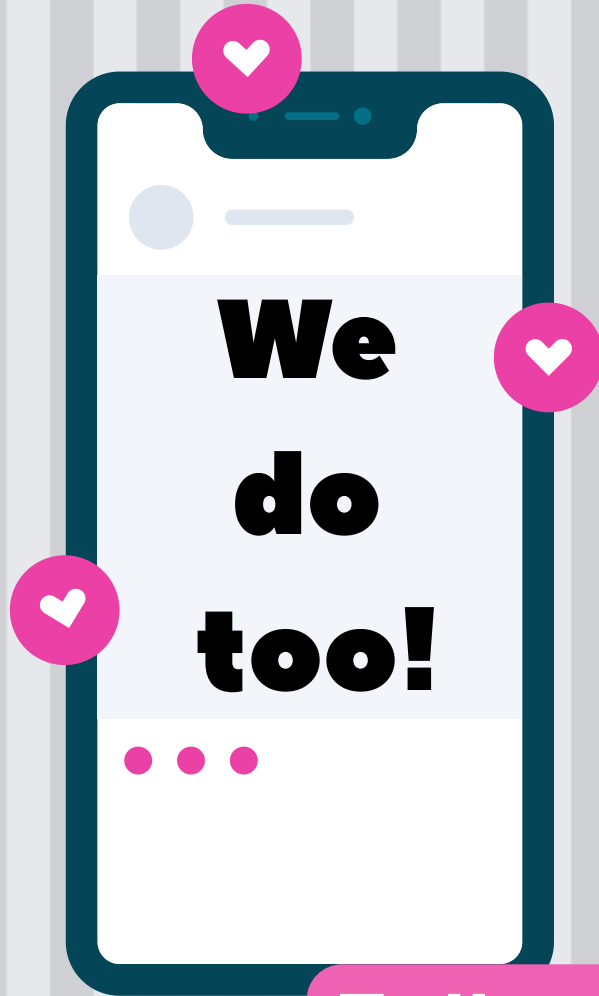
- **How** do I participate?
 - The form takes 3-5 minutes to complete
 - Visit our website and click on "contact register" tab or...

[Click Here](#)

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