The HCU Herald

Featuring...

HCU Hero

Carson from South Carolina

February 2026

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

HCU HERO: CARSON FROM SOUTH CAROLINA

Our Journey from a Marfan Diagnosis to Homocystinuria

For the first time in years, our family felt like we had finally reached a steady place in my son Carson's medical journey. After fifteen years of appointments, specialists, travel, and treatment, we believed we were managing Marfan syndrome well. Carson had been diagnosed at age five with what doctors believed was a spontaneous mutation, since no one else in our family exhibited symptoms. We had done everything we were told to do, and more, to ensure he received the best care possible, no matter the location.

Carson was born long and lean and quickly outgrew his bassinet. He was always the tallest child in swim lessons, mom-and-tot classes, music classes, and at preschool. When he was three, I noticed his chest appeared sunken in while bathing him. Our pediatrician wrote down "pectus excavatum" on a piece of paper and advised me not to Google it.

At age four, Carson was referred to an allergist because his pediatrician suspected asthma. I nearly canceled the appointment, feeling he was too young for testing, but I'm grateful we went. The allergist ruled out asthma, but she began measuring Carson's wingspan and closely examining his chest. After consulting with other doctors in the room, she told us we needed to schedule an echocardiogram immediately because he most likely had Marfan syndrome and could be at risk for a fatal aortic dissection. The news was shocking as we had never heard of Marfan, and we were panic stricken.

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We scheduled the echocardiogram for the next day at a non-children's hospital. The experience was traumatic and unsuccessful. Before we even received results, we were contacted by a geneticist for testing, which prompted us to seek a second opinion at the nearest children's hospital. There, a pediatric cardiologist performed a new echo that came back normal. At the time, a clinical diagnosis of Marfan required five diagnostic criteria; Carson had only one, pectus excavatum, so Marfan was ruled out, and genetic testing was not suggested.

That same year, during a routine kindergarten eye screening, we were told Carson's vision was significantly impaired, and he was referred to an ophthalmologist. That two-hour appointment changed everything. We were told Carson had dislocated lenses, which was something I already knew was considered a hallmark sign of Marfan syndrome after continuing my "motherly instinct" research. We scheduled an appointment with a cardiologist who specialized in Marfan at the same children's hospital and a follow-up echocardiogram showed mild dilation of Carson's aortic root just a short time after his previous normal scan.

At that point, Carson began treatment for Marfan syndrome by starting annual echocardiograms and was prescribed Losartan and Atenolol. Genetic testing was recommended, but it was not covered by insurance and was expensive. We were also told that identifying the mutation would be like "finding a needle in a haystack" and would not change his treatment plan. Given the cost, logistics, and confidence in his clinical diagnosis, especially with the dislocated lenses, we agreed to postpone genetic testing.

In 2017, eight years after his diagnosis, we moved from Texas to South Carolina. Carson's ophthalmologist recommended surgery for his lenses, as glasses were no longer sufficient. To our surprise, the procedure she recommended was only being performed by a handful of surgeons nationwide, and the physician leading the FDA study was located at MUSC.

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She had also trained under him during her residency which led to Carson being approved for the artisan iris claw lens procedure. He underwent surgery just two weeks after our move, and the results were life changing. He no longer needed glasses, and his eyesight improved significantly.

Throughout his follow-ups, both cardiology and ophthalmology, no one questioned his Marfan diagnosis. He had presented an upward displacement of his lenses, pectus excavatum, was tall and thin but “stiff” versus double jointed and had a mild dilation of his aortic root. Diagnostic criteria had evolved, and patients no longer needed five symptoms to be clinically diagnosed as Marfan syndrome was recognized to have a broad and diverse presentation.

In 2020, after a routine cardiology visit, Carson underwent a CT scan to measure his Haller Index. We knew his pectus excavatum had worsened but were told that his organs had accommodated for the space. We were shocked by his measurement of 9.7 which meant there was only a couple of inches of space between his sternum and his spine. We were referred to a surgeon at MUSC for the Nuss procedure, but something didn’t seem right, so I began researching surgeons with extensive experience and recalled learning about cryoablation for pain management at a Marfan conference.

I found a surgeon at the Cleveland Clinic who was a bar consultant, had refined the procedure, and had performed it many times with excellent outcomes.

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Despite the challenges of Covid, we received insurance approval and scheduled the surgery. Carson required three bars, and his sternum fractured during the procedure due to unexpected bone calcification for his age. His surgeon told us it was a complicated case and that the outcome could have been very different without such an experienced team. Four years later, his bars were removed, and once again, we felt we had reached a stable place having addressed nearly every major symptom short of heart surgery.

Carson continued annual cardiology visits at the Cleveland Clinic, along with follow-ups with his surgeon and ophthalmologist, scheduled around his college breaks. This past August, his cardiologist suggested genetic testing as it is now simple, faster, and far more advanced, to which we agreed. Two weeks later, we received a call that changed everything. Carson did not have the FBN1 mutation and therefore did not have Marfan Syndrome. He also tested negative for other connective tissue disorders such as vascular EDS and Loeys-Dietz. Further testing revealed a CBS mutation and a second mutation of uncertain significance. Lab work confirmed elevated homocysteine levels, and the geneticist explained that Carson most likely had homocystinuria.

We were stunned. Despite years of medical care, conferences, specialists, and advocacy, we had never heard of this condition, one that mirrors Marfan in many ways and can be identified with a simple blood test.

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To say our world was turned upside down is an understatement. In the past month alone, we've consulted genetics teams at the Cleveland Clinic, Le Bonheur near Carson's college, and finally a wonderful team at MUSC. My background as a paralegal made me relentless in researching and advocating, which led us to HCU Network America and ultimately back to MUSC, bringing our journey full circle after our move to South Carolina.

Adding to our surprises, genetic testing revealed that both Carson's brother and I are carriers of the CBS mutation, while my husband carries neither mutation. Per the genetic counselor, this suggests Carson's second mutation is spontaneous and not yet classified. He will undergo further genetic testing as his cardiologist believes he may still have an underlying connective tissue disorder contributing to features less common in homocystinuria.

I'm not going to lie; the transition has been overwhelming. In just a few weeks, Carson has had botched labs by the local hospital, started Betaine, B6, and B12, adjusted his cardiac medications, and began restricting protein. The addition of medical formula and further dietary changes are coming, and he hasn't exactly been thrilled. This new diagnosis has brought waves of grief and confusion, especially after feeling so secure in our Marfan journey. Our Marfan doctors and foundation community feel like family, and it was an emotional, possible "last" visit with his treatment team.

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However, the timing has also been a blessing in that Carson has been home for Christmas break, giving us time to adjust, learn, and establish routines together.

After speaking with his Cleveland Clinic treatment team and our Marfan community, this experience has reshaped our perspective and strengthened our commitment to advocacy. Our hope is to raise awareness about early genetic testing, particularly in cases believed to be spontaneous, and to help build connections between the Marfan and HCU communities, so families receive the right diagnosis as early as possible.

In the coming months, we hope to better understand Carson's dietary tolerance with treatment, gain clarity on his genetic results, and build collaboration between the Marfan and HCU communities. Most importantly, I hope to connect Carson with others who understand this journey, so he doesn't feel alone while navigating a life-altering diagnosis during what should be some of the best years of his life.

PANDA EXPRESS X HCUNA Fundraiser

HCU NETWORK AMERICA

Thank You For Supporting HCUNA During Our January Panda Express Fundraiser.

You showed up in Illinois, Texas, Florida, Washington, Idaho, Colorado, and California!

You helped us raise more than our 2025 Panda Express Fundraiser!

PANDA EXPRESS · CHINESE KITCHEN ·

FOOTBALL SQUARE FUNDRAISER

SEATTLE SEAHAWKS VS

FEB 8TH @ 6:30 EST

$20 PER SQUARE 4 CHANCES TO WIN

$200 PAYOUTS 1ST-3RD QUARTER

$400 PAYOUT AT END OF THE GAME

TO PARTICIPATE:
STEP 1: VENMO @TOM-HAWKINS-1
*LAST FOUR DIGITS OF PHONE NUMBER: 1300

STEP 2: EMAIL TOM TO RECIEVE ACCESS TO BOARD!
EMAIL: TMMYHWK09@GMAIL.COM

ALL FUNDS RAISED SUPPORT HCU NETWORK AMERICA'S EDUCATION & OUTREACH PROGRAMS!

WEAR YOUR AWARENESS

Grab your gear!

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

Order by February 5 to receive in time for Rare Disease Day!

February Virtual Meet Up

February 22, at 3 pm CT

REGISTER HERE

RARE DISEASE DAY IS COMING!

RARE DISEASE DAY®

Mark your calendars...

...for February 28, 2026!

One out of every 10 Americans is living with a rare disease.

Rare Disease Day takes place worldwide, typically on or near the last day of February each year, to raise awareness among policymakers and the public about rare diseases and their impact on patients' lives.

How can I participate on Rare Disease Day?

Share your story: Just by sharing your story with others, you're spreading awareness. Whether it's through social media, at school or at work, or in line at the grocery store, each interaction counts!

Wear your awareness: Wear one of your HCU Network America shirts or sweatshirts on Rare Disease Day! And when folks ask you about it, tell them a little bit about YOUR story living with HCU!

Attend an event: There are a number of in-person or virtual events that you can attend! Below are a few options - click each image to check them out and register!

FDA-NIH **RARE DISEASE DAY**
Feb. 27–28, 2025 | #RDDatFDANIH

NORD® National Organization for Rare Disorders

RARE DISEASE DAY ON CAPITOL HILL

Registration opens January 7th, 2026!

Rare Disease Week on Capitol Hill empowers and inspires hundreds of advocates each year. The connections you make during the week will impact rare disease patients for generations to come.

Hosted by the Rare Disease Legislative Advocates (a program of the EveryLife Foundation for Rare Diseases), this multi-day event brings together rare disease advocates from across the country to make their voices heard by their Members of Congress. Participants are educated on policy proposals impacting the rare disease community and provided opportunities to advocate for policy changes directly to their Members of Congress. No matter one's connection to rare disease or their advocacy experience level, all are welcome.

Click here to learn more, and to register to attend!

LET US KNOW!

Planning to attend?

We'd love to connect you with others in our community who will be attending!

MARK YOUR CALENDARS

Advocacy Webinar Series – Collaboration in 2026

Workshop 1: Intro to Advocacy

Advocacy 101: What Advocacy Is—and How You Can Get Involved

Tuesday, March 3, 2026 · 6:00 PM EST
Live virtual webinar (Zoom)

Session Overview

Join HCU Network America, Propionic Acidemia Foundation, and the MSUD Family Support Group, in collaboration with the EveryLife Foundation, for a clear, approachable overview of advocacy. We'll cover the basics, common advocacy pathways, and simple actions you can take to support your community and drive change.

You'll learn

- What advocacy is (and what it isn't)
- Common ways advocates make an impact
- Easy first steps you can take right away
- Practical examples for rare disease communities

Register:

https://us06web.zoom.us/meeting/register/FVKQHTWnR-umU9bh9qp3UQ

Presented by: HCU NETWORK AMERICA | PROPIONIC ACIDEMIA FOUNDATION

PARTNERS: MSUD FAMILY SUPPORT GROUP

In collaboration with: EVERYLIFE FOUNDATION FOR RARE DISEASES

REGISTER NOW

MARK YOUR CALENDARS

Advocacy Webinar Series – Collaboration in 2026

Workshop 2: Storytelling

Advocacy 101: Tell Your Story, Drive Change

Thursday, March 19, 2026 · 6:00 PM EST
Live virtual webinar (Zoom)

Session Overview

Your story is a powerful advocacy tool. Join HCU Network America, Propionic Acidemia Foundation, and the MSUD Family Support Group, in collaboration with the EveryLife Foundation, to learn simple storytelling frameworks, key message elements, tips for sharing your story with policymakers and others with confidence, and practice with your fellow advocates.

You'll learn

- How to shape your story for impact
- Key message elements that "stick"
- Tips for sharing with policymakers and partners
- How to communicate with confidence

Register:

https://us06web.zoom.us/meeting/register/zkED4UJFQBiklyF5m7hyqQ

Presented by: HCU NETWORK AMERICA | PROPIONIC ACIDEMIA FOUNDATION | MSUD FAMILY SUPPORT GROUP

PARTNERS

In collaboration with: EVERYLIFE FOUNDATION FOR RARE DISEASES

REGISTER NOW

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!

Click here to learn more and to start your application!

The application deadline is March 15, 2025.

Click Here

HCU NETWORK AMERICA

MEET OUR BOARD MEMBERS

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MEET OUR
BOARD MEMBERS

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WELCOME TO OUR NEW

TREASURER

Benjamin Lewis

Diagnosed with classical HCU at birth, Ben is a lifelong patient advocate using his voice to drive progress for the HCU and rare disease communities. As a board member, he brings both lived experience and strategic leadership to our mission.

CLICK HERE

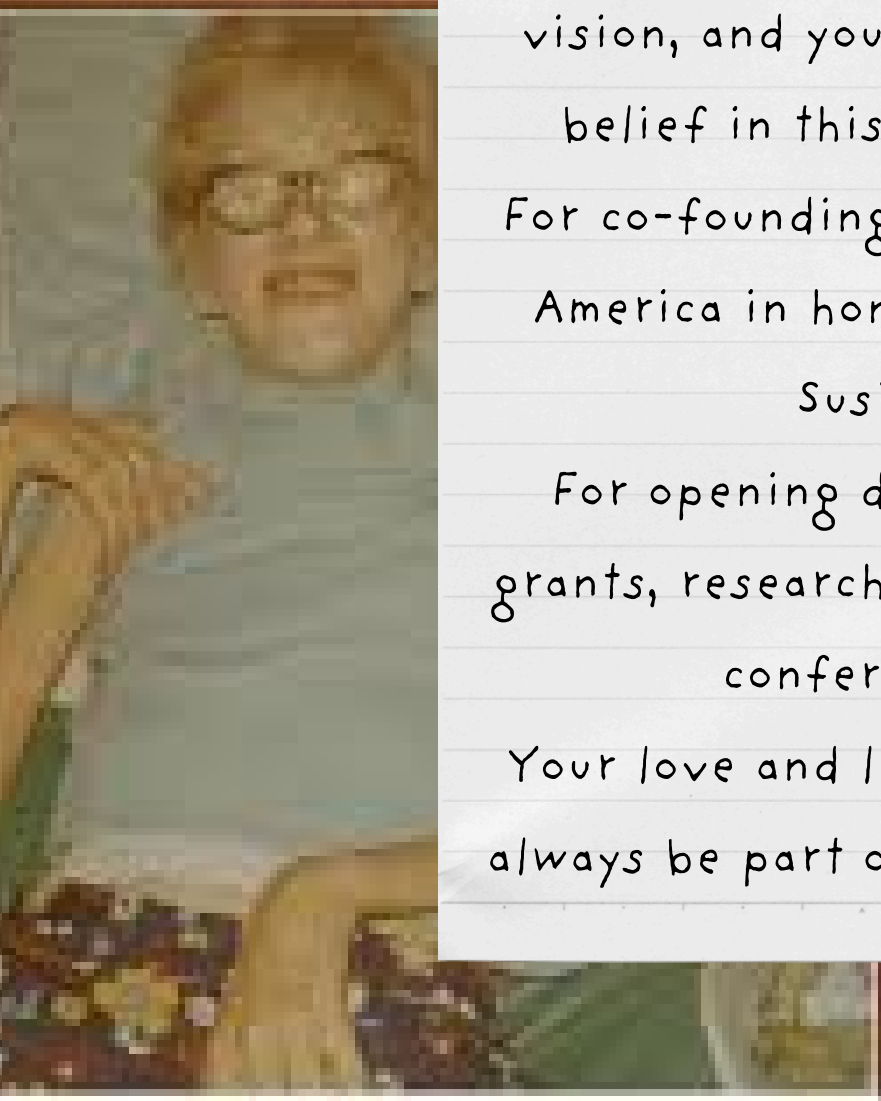
Thank You

Margie, for your heart, your vision, and your unwavering belief in this community.

For co-founding HCU Network America in honor of Judy & Susie.

For opening doors through grants, research, and our first conference.

Your love and leadership will always be part of who we are.



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

CALL FOR ABSTRACTS SUBMISSION DEADLINE APRIL 30th, 2026

July 10-12, 2026

Posters on:

- Natural history
- Novel therapies
- Treatment
- Diagnostics & Testing
- Other

Abstract Submission Link

For questions, email research@hcunetworkamerica.org

Please note: accepted abstract authors will need to register as a medical professional or industry

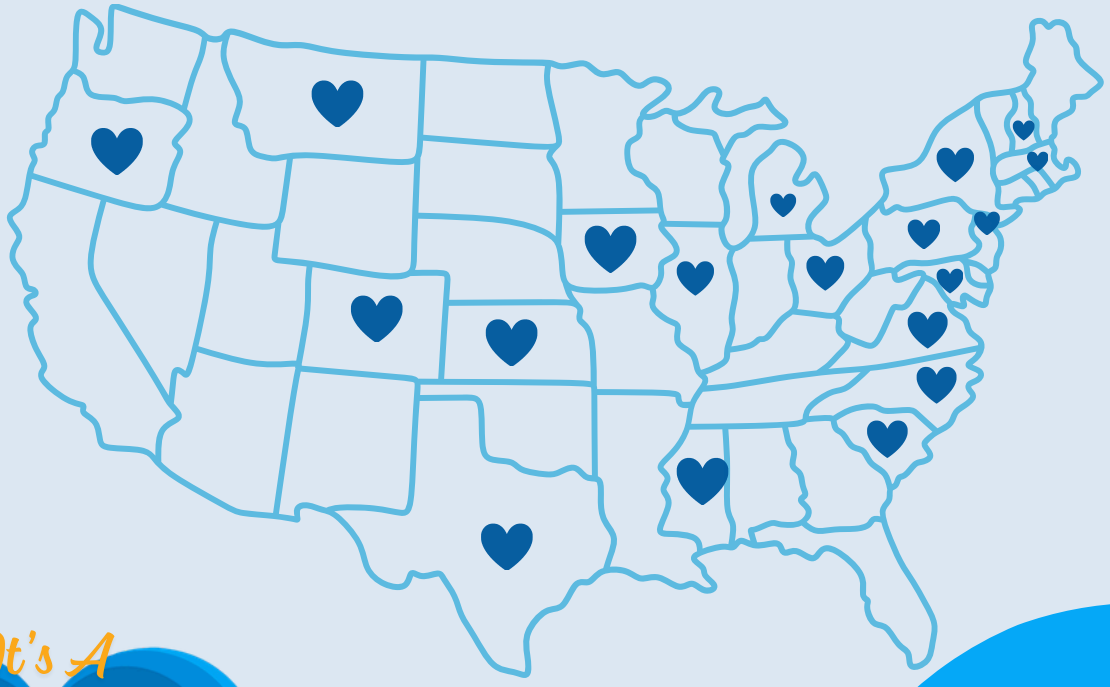
2026 CONFERENCE

Attendees From Everywhere

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

2026 CONFERENCE

Attendees From Everywhere



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

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In The City of Brotherly Love

July 10-12, 2026
Philadelphia, PA

- Patient Focused Panels
- Breakout Sessions
- KidsZone and Teen Zone
- Community Building Opportunities

- Dedicated Science Day for Families and Professionals
- Scientific Poster Session
- HCU Hero Award Banquet
- Networking Opportunities

Patient & Family Conference

REGISTER NOW

HCU NETWORK AMERICA

Classical HCU | Cobalamin Disorders | Severe MTHFR

Rare Study Comparisons

NORD National Organization for Rare Disorders | LIVING RARE Study

RARE-X A Research Program of Global Genes

LIVING RARE Study	RARE-X
Participants can have any type of rare disease	All Participants have any form of Homocystinuria
	You own your data and consent to how it is used
Surveys focus on experiences and challenges of people impacted by rare disease	Surveys focus on symptoms and quality of life related to HCU
Show the significant unmet needs of the rare disease community	Updating surveys yearly builds natural history data
Transform lived experiences into facts that will illustrate the impact of rare disease for key decision makers	Data is used to advocate for better management of HCU and inform clincial trial Development for new treatments
Leverage the strength of collective voices in advocating for a better future for all impacted by rare diseases	Contribute to disease understanding which paves the way for a brighter future of HCU patients

HCU NETWORK AMERICA

Your time is valuable. Informing yourself on different studies available will help you decide where to dedicate your time and energy.

HOMOCYSTINURIAS DATA COLLECTION PROGRAM

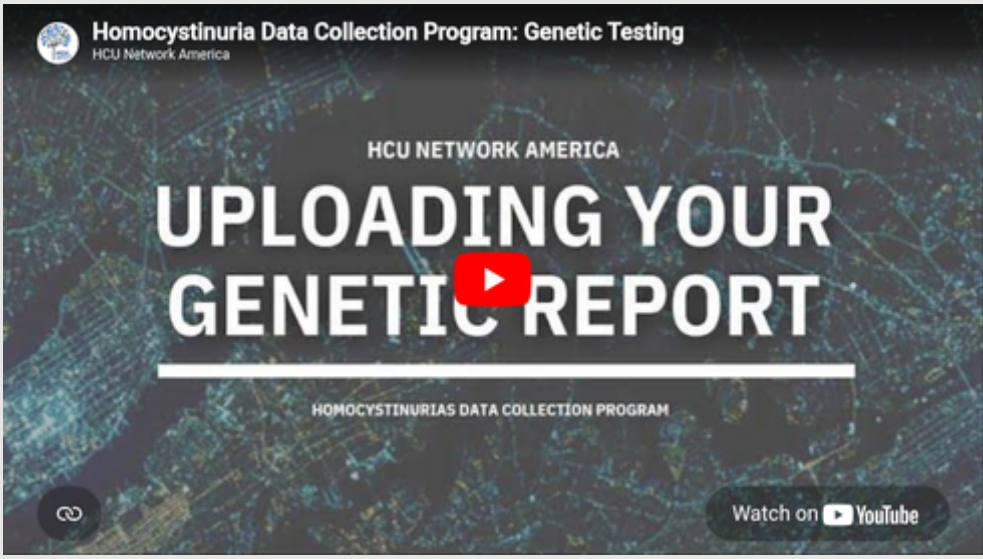
POWERED BY RARE-X

UPLOADING YOUR GENETIC REPORT

Did you know a genetics report can help build a better understanding of homocystinuria?

Watch to find out:

- What is genetic testing?
- The components of a genetic report
- How I upload it into Rare-X
- Benefits of a genetic report
- Ways to get tested
- What you should expect



Upload your genetics report:
https://rare-x.org/homocystinuria/

CANPKU+ CANPKU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

METHIONINE IN HOMOCYSTINURA

Guideline Recommendations

Monitoring methionineinadditionto homocysteineiscrucialtoassess the effectiveness of treatment and risk of complications

Diagnosis	Recommended Plasma Methionine Level	Reason
CBS Deficiency (Classical Homocystinuria)	Less than 1000umol/L	Decreases risk of cerebral edema and encephalopathy
Remethylation Disorders (Cobalamin and Severe MTHFR)	10-40 µmol/L Within Normal Limits	Ensure availability of methyl donor reactions

Important Considerations

Betaine treatment must be managed appropriately to ensure plasma methionine levels remain in the recommended range

Regular labs are recommended for close monitoring of plasma methionine along with plasma homocysteine levels
Childhood (Every 3-4 months), Adolescence & Adulthood (Every 6 months)

Low methionine in patients with remethylation disorders increases risk of developmental delay, seizures and intellectual disability.

Suggested Laboratory Findings-CBS Deficiency

Following ahigh homocysteine lab result,plasmamethioninelevelswithinthe following ranges, could suggest a diagnosis of inherited homocystinuria.

	Childhood-Onset Multisystem Disease	Adult-Onset Thromboembolic Disease	Normal Range
Plasma Methionine Concentration	**200-1,500 µmol/L**	**>50 µmol/L**	**10-40 µmol/L**

https://www.ncbi.nlm.nih.gov/books/NBK1524/ https://jcp.bmj.com/content/75/11/744.long

HCU NETWORK AMERICA

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!

The latest episode

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!

Ben welcomes Melanie to the table!

Last time we heard from Melanie, she was sharing her son's journey to a late diagnosis. Tune in for an update on how he's doing and how the family has adapted after learning that Masen has Classical HCU.

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

CUSTOMIZE YOUR KIT FOR FREE!

At HCU Network America, we believe that one of the most important steps to empowering patients and caregivers is giving them the support and tools needed to succeed! We know that a new diagnosis can be overwhelming and riddled with concerns and questions. To us, one way to combat those feelings, and give you the confidence you need, is by providing you with one-on-one support, educational resources, and practical tools, such as scales, cooler bags, and more! Our request for a kit survey allows you the opportunity to request a one-on-one introductory call (with more opportunities to connect), and then a customized kit to the patient's needs. Don't want a call or a Zoom? That's fine too - we are happy to send you the customized kit.

Request your kit now - **https://www.surveymonkey.com/r/HCUKitSurvey**

**Kits can only be sent to patients in the continental US. However, we are happy to connect virtually and share the educational materials with you via weblinks*!

Contact Register

What?

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

Why?

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

How?

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

Click Here

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HCU Network America

@HCUAmerica