

The HCU Herald

Featuring...



HCU Hero
Madison from New Jersey



June 2024



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

HCU HERO: MADISON FROM NEW JERSEY

My name is Madison Manno and I recently turned 19 in March 2024. I was diagnosed at birth with classical homocystinuria. My older brother, who was born in 1991, also has classical HCU, but he wasn't diagnosed until he was four and a half years old. At age eight, both lenses in my brother's eyes became detached about a month apart from one another. This was a severe side effect of homocystinuria. He was left needing a very unique prescription. He had glasses and contact lenses that were made just for him, and the lenses were incredibly thick.

My mother always wanted four children but between taking care of most of my cousins and my two older siblings (including one with homocystinuria that required extra attention), they just didn't have the time to add two more children into the mix. But in 2004, they decided to have a third child and in March 2005, I was born. When my mother found out she was pregnant with me, her doctors already knew about my brother's diagnosis of homocystinuria, so as soon as I was born, I was rushed to Children's Hospital of Philadelphia to be tested for homocystinuria. It was there that I was diagnosed and started a special infant formula.

When I was younger it was a lot easier for my parents to be in control of when I was taking my medicine and restricting my diet. The older I got the more my peers started noticing that I would pack lunch a lot or I didn't have chicken nuggets in my lunch like most of the other kids did. At the time, they didn't think much about it, and neither did I. I think I was in first or second grade when I had the realization that I was "different" from my peers, although I didn't yet understand what it was that made me different. I would get pulled out of class for "special time" with different teachers, and when the ice cream truck would come visit us on the last



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day of school, I had to get a popsicle and all the other kids got to have ice cream on a cone with sprinkles. It wasn't until around third grade that I realized I had something "wrong" with me that made me different from my peers. I remember my mother would mix my HCU cooler pouches in the morning in a cup and I didn't like the taste of them. We had sinks in our classrooms so every morning when I'd get into class, I would pour the liquid into the sink, and down the drain it would go. My mom would always be so proud of me for "drinking" the HCU coolers, but I secretly hated having to drink it. I even had a special cup to try and encourage me to drink it, but with time this "special" cup didn't really seem so special anymore. My peers would say that it smelled bad and it made me insecure to drink it. I had the fear that if I drank it, it would make my breath smell, so every morning I poured it out and lied about drinking the HCU cooler; same thing with my Cystadane powder. It had a horrible stench, and I became so fearful that it would have the same effect on me, so I also started to pour my medicine down the drain when my parents weren't weren't paying attention. I think I eventually got caught and my parents were extremely upset. When I entered the fifth grade my parents let me have a little more freedom while getting myself ready in the morning. This meant it was my own responsibility to take my medicine in the morning and before bed, along with everything else that I needed to do to prepare for the day. I still didn't take medicine as regularly as I should have. This lasted until about the end of my freshman year of high school.

I loved everything about high school. I hated my elementary/middle school, mainly because of my peers. They constantly picked on me for being so skinny and tall. They always found ways to criticize me and make me feel less than, whether it was about the way I looked, what I ate that day at lunch, or how I wasn't as "smart" as them. I remember having a really hard time socially and educationally due to the severity of being picked on by my peers. All of my friends were in the two clubs that were in our school: student council and safety patrol. I tried for four years to get into the clubs, but I was never accepted. Looking back, I think a lot of it was due to the fact that I had homocystinuria. I think the teachers thought that I wasn't as capable as other

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students, mainly because of my IEP and because of having my brother as a student when he was younger. My brother struggled a tremendous amount during his school years, and I think because they saw how much he struggled, they thought my experience would be the same. During sixth grade, I was contemplating asking my parents if I could switch to another class because I was struggling so much socially and somehow it got back to my teacher. I remember she took me out of class to talk in the hallway and she asked me if the rumor was true. I said, "Yes, but I haven't talked to my parents about it. I was just thinking about it." I remember she said, "If you switch classes, you won't ever be able to do it. You can't do it."

...my entire life I was told that I wasn't capable of being in the "smart class." He was the one who truly made me realize I was capable of doing so much more than I thought I could.

When I went into high school I kind of let go of my diet because I wanted to fit in. I hated the way my previous set of peers treated me, so I wanted this time around to be different. I was a bit nervous because I didn't want to be known as the "stupid kid," like I was in my other school. It wasn't until my sophomore year that I sat down with my guidance counselor and he told me he wanted me to try a couple of honors classes. I was so scared because my entire life I was told that I wasn't capable of being in the "smart class." He was the one who truly made me realize I was capable of doing so much more than I thought I could. If I remember correctly, I finished that school year with straight A's. I graduated from high school as an honors student - I was in many honors and AP classes, I was the president/vice president of so many clubs/honor societies during my high school career.

In March 2020, the world shut down due to COVID. I was devastated because at this point, I had fallen in love with school and my social life. I actually had friends and always had something to do. It completely destroyed me, not being able to socialize and be around others. During COVID I got really into health and fitness. This had both positive and negative effects on me in the long run. I definitely didn't start my health

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and fitness journey in the most positive way, but I can say today that I have learned so much from my mistakes in the past, and I am still continuing to learn better ways to improve my health today. Your body is what makes you, you. You shouldn't feel ashamed for fueling yourself. Everyone needs a different amount to fuel their energy. For the longest time I struggled with what to put into my body because my levels were never good when I would get bloodwork done, but I also struggled because I started gaining weight. I eventually figured out what was best for me and my body. When shops and restaurants finally started opening up again, I got a membership at a gym. The gym has helped me in so many ways in accepting my body and learning to be easier on myself than I was in the past. Also, because of the gym, I started to take my medication more seriously. I wouldn't say at that point in time that my diet was that much better, but the gym definitely helped me start to improve it.

This past year when I found HCU Network America it helped my HCU journey tremendously. Seeing so many other individuals of all ages going through exactly what I am on a daily basis is comforting because it reminds me that I'm not alone on this journey. It's all a learning experience of trial and error and learning what works best for you and your lifestyle.



Almost a year ago, I remember seeing HCU Network America repost a video on social media of another patient. Her name was Gabbi, and she had made a video of a day in her life as a patient with homocystinuria, and for me, that was life-changing. Sometimes when I'm having a hard day I think about that video and Gabbi, or I even go back and watch it. It reminds me of how strong I am despite all of the trials that I've had to face living with this disease. Gabbi and I have recently connected on Instagram, and it feels so nice to be connected to someone else around my age who understand what it's like to live with HCU.

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In February 2023 I competed in my first pageant and in April of that same year, I won my first pageant (this was only my third time competing) and was crowned the title of Miss Tri-City 2023. As Miss Tri-City, I promoted HCU as my platform to promote not only the awareness of HCU, but also to share my story with others. I shared how I have been able to overcome so many obstacles despite my rare disease. My entire life I have been told that I wouldn't be able to achieve so many things. Those words have stuck with me forever in my educational career, but here I am today writing about my story and sharing it with so many others to read.



I just finished my first year of college. In May 2025, I will graduate with my associate's degree in liberal arts elementary/secondary education AA. I'll continue my education to obtain my bachelor's degree in music education. My goal is to become a music teacher and hopefully teach high school students. I hope to be a positive influence towards others, not only to patients with HCU, but to all people who have ever been told they can't do something, because you most certainly can. We can ALL achieve great things. It may be harder for some than others, but you shouldn't let that stop you. It just makes the end goal that much more rewarding.

[Click here to read Madison's story on our website](#)

We need Patient Stories!



BY SHARING YOUR STORY, YOU CAN....

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

Ready to share your story?

Send us a message on social media or email us!

info@hcunetworkamerica.org



JOIN OUR FUNDRAISING TEAM

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

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

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



To join, email Dbartke@hcunetworkamerica.org

World Homocystinurias Awareness Day – recap!

World Homocystinurias Awareness Day is an initiative of patient organizations and consortia globally who have a focus on the homocystinurias. The main objective of World Homocystinurias Awareness Day is to raise global awareness of the homocystinurias and their impact on patients' lives.

WORLD
HOMOCYSTINURIAS
AWARENESS DAY 2024



to all who participated by spreading awareness and by participating in our Raffle Fundraiser or making a Donation!

We were able to raise \$3,256 for our Education & Outreach programs!

Raffle Winners

Jackie Piccini



Melanie Colter



Liz Carter



Ilyse Brainin



Linda Conard



Steve Holley



Carson Talbert



Danae Bartke



Cynthia Kushnier



A special thank you to Dean Seppelfrick, Heather Eckman, Austin's Violin Shop, Kendra Scott Jewelry and the Bartke Family providing our awesome prizes!



HCU HEROES RACE FOR RESEARCH THIS SEPTEMBER WALK | RUN | RIDE



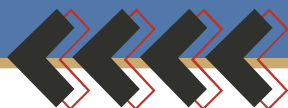
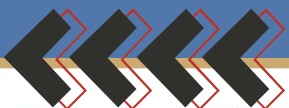
Cystadane; the last FDA approved drug for the homocystinurias (HCU) was approved in 1996. That was 28 years ago!



Currently is only one clinical trial in progress for classical HCU, but there are none in progress for cobalamin disorders and Severe MTHFR.



In collaboration with other organizations, HCU Network America has issued 7 research grants.



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



<https://bit.ly/HCURace24>



Pricing

- \$5 - no shirt or medal, participate and fundraise!
- \$20 Medal or shirt only
- \$30 Both!

Conference Registration Deadlines approaching!

- Last day for hotel discounted rate: June 4
- Last day for conference registration: June 5

10 reasons to attend the 2024 HCU Network America Family Conference



Register here: <https://bit.ly/hcuconference2024>

Registration:

[TICKETSIGNUP.IO/TICKETEVENT/HCUCONFERENCE](https://ticketsignup.io/ticketevent/hcuconference)

MOVING MOUNTAINS

Classical HCU | Cobalamin Disorders | Severe MTHFR

Patient-Family Conference

Aurora, Colorado
June 29-30, 2024



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



UPCOMING EVENTS



HCU COMMUNITY VIRTUAL MEETUP

Monday, July 15, 2024 | 8 pm ET

We hope you'll join us!

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



<https://bit.ly/summer-meetup>




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.



Clinical trial & research discussions

What topics would you like to discuss in regards to clinical trials...


Please comment 9 →



Do you have any HCU Diet & Nutrition tips or ideas to share?

Diet and nutrition can be an important aspect of HCU management. How...

Please comment 4 →



Homocystinuria - Data Collection Program

The HCU Networks have partnered with RARE-X to drive a Data Collection...

Please comment 1 →



Join the conversation!
<https://hcuenetwork.org/>

Incase you missed it...



The HC&U Podcast is back!!!

HC&U is a podcast about Homocystinuria, sponsored by HCU Network America and hosted by Ben & Lindsey.

Meet your hosts!



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



To Listen:

<https://hcunetworkamerica.org/hcu-podcast/>
or click below on your favorite option!



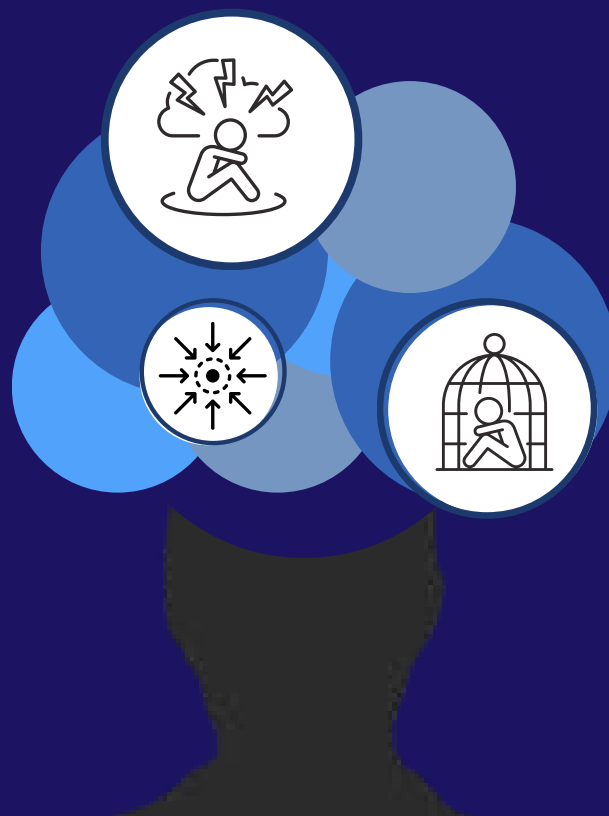
The latest episode



In this episode, Ben welcomes Alex Orange Drink to the table! Alex is a musician/songwriter who lives with Classical HCU.

Ben & Alex discuss the mental health challenges that growing up with a rare disorder presents and how they've coped over the years. They also leave us with some great advice for the younger generation of HCU patients.

In a study from a 2019 publication, "Revising the psychiatric phenotype of homocystinuria", 16 of the 25 patients in the sample (64%) reported psychiatric symptoms, including a high prevalence of both anxiety (32%) and depression (32%). Deficit–hyperactivity disorder (ADHD), oppositional defiant disorder (ODD), mood swings, hallucinations, and suicidal thoughts have also been reported.



Complete the Behavior Survey

homocystinuria.rare-x.org

Samantha's "Beefy" Mac



Makes about 3 servings | 1 serving = 2 oz | 1.1 grams protein per serving

Ingredients:

- 112 g Aproten Penne, dry
- 2 TBSP Olive Oil
- 2 TBSP Diced Raw Onions
- 1 clove(s) Garlic, minced
- 1/4 c Universal Ground Beef
- 7 g Cambrooke Foods Burger Patty Mix
- 1/4 tsp Smoked Paprika
- 1 tsp Garlic Powder
- 1/2 tsp Onion Powder
- 1 TBSP Shake 'N Cheese
- 1/2 c Canned tomato sauce
- 1 oz. Violife Just Like Cheddar Shreds
- 2 TBSP Rice milk

Directions:

1. Cook pasta according to directions on the package.
2. While the pasta cooks, heat the olive oil in a medium skillet over medium heat. Add the onions and garlic and cook for two minutes, stirring often to prevent the garlic from burning. Add the ground "beef" to the skillet and the patty mix. Cook until the "beef" mixture thickens. You can add a little more oil if it sticks a little. Next add the spices, Shake n Cheese, tomato sauce, and rice milk. Cook until thickened. Add the shredded cheese and cook until melted.
3. Drain pasta and rinse well. Add to the sauce mixture and stir to combine. Add salt and pepper as needed. Serve immediately.

Note: If you do not have Shake n' Cheese you can use another powdered cheese like Molly Mcbutter (or omit). If you do not have Cambrooke's Patty Mix, you can use 1.5 tsp wheat starch.

Tip: Cook the pasta in vegetable or chicken broth for additional flavor for the pasta.

UPCOMING EVENTS



**GREAT OAKS
RETREAT CENTER**

ILLINOIS LOW PROTEIN FAMILY CAMP

REGISTER NOW!



Check-in: 5:30 pm, August 9, 2024 | Check-out: 10 am, August 11, 2024

Great Oaks Retreat Center
1380 County Rd 900 N, Lacon, IL 61540



LODGING

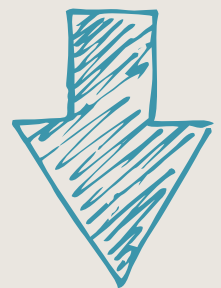
Lodging is included. Please bring your own pillows and linens - blankets or sleeping bags. Beds are bunk beds in a large room.



WHAT TO BRING:

- Formula
- Sleeping bag or linens
- Favorite snacks
- Towel
- Bathing suite
- Sunscreen
- Bug spray
- Clothes to camp in
- Closed toe shoes
- Shampoo & soap
- Tooth paste & toothbrush
- Camera
- Phone charger

REGISTER NOW!



<https://bit.ly/3TKWdJc>

**FRIDAY
08/09**

Potluck Dinner
Arts and crafts
Bonfire and smores

**SATURDAY
08/10**

Breakfast
Tie Dye
Group Photo
Archery

Open swim Boating Fishing
Lunch
Ziplining/high ropes

Dinner
Ghost in the Graveyard
Bonfire and Smores

**SUNDAY
08/11**

Breakfast
Closing Activity
Clean and pack up

**REGISTER
NOW!**



Now
Enrolling

Acappella

Sponsor: Traverre Therapeutics

Study type: Natural History (no investigational medicine given)

Study duration: About 6.5 years

Goal: To learn more about classical HCU & the course of the disease

TO QUALIFY*	AGE OF PARTICIPANTS	DETAILS
Diagnosis of Homocystinuria due to CBS Deficiency (Classical Homocystinuria)	Currently enrolling 1 to 4 years old	The study will include three key stages (screening, enrollment, and observational follow-up) and will last approximately 6.5 years.

**Your child will need to meet all other study criteria to take part in the ACAPPELLA Study.*



Study Locations

United States: Colorado, Washington DC, Georgia, Pennsylvania

Countries outside of the US: Ireland and Qatar



Approximately 150 people

aged between 1 and 65 will participate at sites in the US, Europe and other countries around the world.



The ACAPPELLA Study has already enrolled 100 adults and children over 5 years old, and is now looking for children aged 1 to 4 years old to take part.



You may be able to receive payment for time and travel if your child participates in this study.

Talk to your doctor and family members about your child joining the ACAPPELLA study.

Sites are open and currently enrolling participants. For the most current information about the ACAPPELLA Study and to see additional eligibility criteria, please visit:

<https://www.clinicaltrials.gov/study/NCT02998710>

If you have any questions, please email:

medinfo@traverre.com



For more information, please scan the QR code or visit:

www.hcuconnection.com

Sign our NBS Screening Petition!



Give Hope,
Help Save Lives

#NewbornScreeningSavesLives

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



Click [here](#) to sign the petition!

UPDATE

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



CLASSICAL HOMOCYSTINURIA MEDICAL ASSISTANCE PROGRAM

What is the purpose of this program?

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

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

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

**MEDICAL
ASSISTANCE**



Who is eligible to apply?

This program is designed to help patients who:

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



What is the application process?

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

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

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

NORD is Here for You

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

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

Alone we are rare. Together we are strong.®

How do I get more information and apply?

Contact NORD's Classical HCU Medical Assistance Program

Monday-Thursday 8:30am – 7:00pm ET

Friday 8:30 am – 6:00pm ET

☎ 203-616-4327

📠 203-635-4163

✉ hcu@rarediseases.org

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



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

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

What assistance does NORD provide?

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

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

Awards are issued for a calendar year.

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

How does NORD demonstrate compliance with regulations required of charities?

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

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!*

Stay connected: Join our Contact Register!

What is a contact register?

- a secured private survey that allows you to share information on you or your family member with HCU with us. (general contact info, diagnosis, etc)
- kept confidential and will not be shared unless you permit us



Why join?

- subscribe to our monthly newsletter and other communications
- identify other patients in your state and request their contact information
- access information posted over time that can only be shared with the patient community
- helps us plan events and inform the development of resources and educational tools
- supports our advocacy efforts and enables us to have informed conversations with doctors, pharmaceutical companies, state newborn screening labs, and lawmakers.

I want to participate! What's next?

You can find the form either by visiting our website and clicking on the "Contact Register" tab, or you can fill it out by going directly to:

<https://www.surveymonkey.com/r/HCUContact>

H PE CONNECTS US



**FOLLOW
US**

