The HCU Herald

Featuring...

2025

¡en español!
(translation to English included)

HCU Hero Enelcita from Ny

January 2025

HCU NETWORK AMERICA

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

HCU HERO: ENELCITA DE NUEVA YORK

hola

We're excited to share our first Spanish-language HCU Hero story!
The English translation will follow on the subsequent pages.

Me llamo Yasenia Castro y mi familia y yo somos de Honduras. Soy la hermana de Enelcita Castro, quien fue diagnosticada con homocistinuria clásica a los 36 años.

Enelcita fue una niña bien alentada, pero toda su vida tuvimos un diagnóstico equivocado. Cuando estaba pequeña, ella tuvo sarampión y le dio fuerte y de allí los doctores que la vieron dijeron que por causa de la fiebre, ella había sufrido retraso en el desarrollo. Ella dejó de caminar un tiempo y después volvió a caminar más o menos a sus tres años. En Honduras las evaluaciones que le hacían a Enelcita le decían a mi mamá que ella mentalmente tenía unos 2 años y 3 meses más o menos. Ella nunca fue a la escuela porque en mi país no hay escuelas especializadas.

En el 2012, Enelcita sufrió de una fractura de cadera, la cual tuvieron que reemplazar. La recuperación fue lenta pero ella fue tan fuerte que por si sola volvió a caminar.

A los 31 años, Enelcita viajó a Estados Unidos para buscar respuestas a todos los problemas médicos que había atravesado. Los doctores que visitó no sabían tampoco la razón de sus síntomas, ya que la información que teníamos sobre su enfermedad era la que se nos había comunicado desde que ella era una bebé. Se descubrió que tenía coágulos de sangre en las piernas, pero los doctores los atribuyeron a la cirugía de cadera que tuvo, ya que estuvo inmóvil en la cama por un tiempo. Empezó a tomar medicamentos para la sangre. La revisó el oftalmólogo y determinó que su cristalino se había luxado, y nos dijeron que pudo

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haber sido debido a una caída. También fue referida con una psiquiatra, la cual le recetó medicamentos para mantenerla calmada ya que su comportamiento variaba de enojada a tranquila. No funcionaban muy bien y la mantenían dormida por ratos durante el día. Por la noche no dormía bien; daba dormidas de unos minutos y ya volvía su energía. La música se convirtió en una aliada para nosotros ya que ella se calmaba por ratos. Para nosotros, la familia, era muy difícil. Nos turnábamos para cuidar a Enelcita; nuestra mamá la cuidaba más por la noche y yo durante el día.

Fue por esta época donde le descubrieron piedras en la vesícula, lo cual nosotros no sabíamos ya que ella no se comunica verbalmente. Siempre teníamos que adivinar si ella estaba con algún dolor ya que fiebres no presentaba y ella, entre todo, era bien alentada. Le suspendieron los medicamentos de la sangre porque ya no tenía coágulos en las piernas.

A finales del 2019 la llevamos a emergencia. No teníamos una razón específica, solo que no era ella misma. Fue allí donde le hicieron todos los exámenes y su presión no era normal. Cuando la visitaron los doctores y hablaron conmigo a ella la llamaban la Avenger, pero realmente yo pensé que lo decían por su enfermedad, que se comportaba bien. Y no fue hasta que ya nos dijeron lo que habían encontrado en los exámenes que le habían realizado que realmente lo entendí. Los exámenes identificaron problemas de circulación de las venas que llevaban sangre a las piernas, coágulo en la vena principal aorta, y osteoporosis. Aun así no sabíamos que era homocistinuria.

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No fue hasta principios del 2020 que tuvo Enelcita una cita de seguimiento y conocimos al doctor Hussan Alkaissi. Estaba haciendo su residencia en el hospital Downstate y la revisó y nos escuchó. Cuando terminó me mencionó la homocistinuria, ya que él la había visto anteriormente, pero me dijo "No te digo que eso es lo que tiene; pero voy hacerle unos exámenes de sangre." Cuando los resultados le llegaron, él personalmente me llamó y me confirmó que mi hermana si tenía homocistinuria. Me brindó información y me habló de videos en YouTube. El doctor hizo lo posible para encontrar un doctor de genética que la atendiera ya que en Nueva York no tenía muchas opciones. Luego encontró a la Dr Melissa P. Wasserstein de genética pediátrica que aceptó verla y allí fue que ya supimos que sus niveles de homocisteína los tenía a 550, si recuerdo bien.

Me acuerdo muy bien cuando el doctor me explicó sobre esta condición. Lloré porque me sentía tan culpable porque yo alimentaba a mi hermana con mucha proteína. Yo la miraba como tan flaquita y pensaba que tenía que ganar masa muscular. Al no saber qué era lo que ella en realidad tenía, ella comía de todo; no llevaba ninguna dieta en especial.

Desde entonces busque videos para aprender más sobre la HCU y empecé a seguir la página de HCU Network America. Un día asistí a un Zoom y escuché personas compartiendo sus historias. Empecé a escuchar las historias de personas con la condición y como fueron sus síntomas. Fue allí donde me di cuenta de que los síntomas que Enelcita tuvo de niña eran los mismos, solo que con el diagnóstico incorrecto. Fue gracias a esos testimonios y a una pregunta que hice cuando entré a ese Zoom que obtuve la ayuda necesaria para entender mucho mejor

“Empecé a escuchar las historias de personas con la condición y como fueron sus síntomas. Fue allí donde me di cuenta de que los síntomas que Enelcita tuvo de niña eran los mismos, solo que con el diagnóstico incorrecto.”

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a mi hermana. Mi mamá empezó a comprender mejor porque Enelcita no dormía bien, y la razón por su comportamiento. A pesar de que somos 4 hermanos, Enelcita es la única en la familia con esta condición.

El diagnóstico de Enelcita cambió por completo a la familia, ya que ahora teníamos que seguir con un plan de alimentación, administrar la fórmula de aminoácidos, Betaina y el resto de las medicinas que ella necesitaba para su condición. Mi hija y yo tomamos el control de la alimentación y sus medicamentos ya que nosotras hablamos inglés y nos entendimos con los doctores. Mi mamá obtenía la información de nosotras ya que ella no hablaba inglés. Fue un gran alivio saber que gracias a la dieta y los medicamentos, empezamos a ver cambios en Enelcita con el tiempo. Sus niveles de homocisteína fueron bajando y hoy en día están a 90. Su desarrollo ha sido grande y ha retomado el aprendizaje. Ahora ella pone mucha más atención, como un niño creciendo. Ella baila, canta, y le gusta pintar. Pide lo que quiere ver en la tv y está diciendo palabras en inglés lo que nos ha sorprendido mucho. Ella no habla oraciones completas, pero la entendemos.

En el 2021 mi mamá falleció y ahora Enelcita es mi responsabilidad completamente. Pero gracias a la ayuda de mi hija, hemos logrado mucho avance con Enelcita a pesar que no tuvo un diagnóstico a temprana edad. Ahora estamos disfrutando el proceso y nos alegra la vida. Desde que Enelcita nació, empezó su vida con drama: nació en el carro, no esperó llegar al hospital. Desde su nacimiento brilló y ella es nuestro milagro. Es por eso que quiero compartir su historia. No sé cómo sobrevivió a tantos años sin un diagnóstico, como su cuerpo aguantó tanto, y cómo se desarrollaron nuevas venas para que le llegara sangre a las piernas. Así como los doctores la llamaban "una avenger", para nosotros es un milagro.

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Muchas veces en el pasado tuvimos discriminacion por parte de algunos médicos pero siempre seguimos buscando nuevos. Gracias a Dios llegó el doctor que no le tuvo miedo y que la atendió con respeto. Reconoció los síntomas de mi hermana y gracias a él, finalmente pudimos obtener el diagnóstico correcto después de tantos años. Ahora él terminó su residencia, se fue y dejó a otro doctor encargado de mi hermana. Aunque él no esté allí, el nuevo doctor, Shiang Lin comparte las actualizaciones de Enelcita con él.

Si puedo ayudar a otros, sería por medio de la historia de mi hermana; que si no tienen un diagnóstico que no se cansen de buscar otras opiniones, que el doctor adecuado llegará con las respuestas correctas. Enelcita es nuestra heroína, nuestro milagro y si sigue con nosotros es por un propósito. Mi consejo para las personas que tienen un familiar con HCU es que se llenen de paciencia. La vida no siempre es fácil, pero vale la pena el resultado.

Haz clic aquí para leer la historia de Enelcita y las de otros pacientes en nuestro sitio web!

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Enelcita's story: English translation

My name is Yasenia Castro and my family and I are from Honduras. I am the sister of Enelcita Castro who was diagnosed with classical homocystinuria at the age of 36.

As a child Enelcita was relatively healthy, but her entire life she had the wrong diagnosis. When she was little, she had a severe case of the measles. The doctors who saw her said that because of the fever, she had a developmental delay. She stopped walking for a while and then started walking again at about the age of 3. In Honduras, Enelcita was evaluated, and my mother was told that she was mentally about 2 years and 3 months old. She never went to school because in my country there are no specialized schools.

In 2012, she suffered a broken hip, which had to be replaced. Her recovery was slow, but she was so strong, and she walked again on her own.

At age 31, Enelcita traveled to the United States to seek answers to all the medical problems she had been going through. The doctors she visited did not know the reason for her symptoms either, since the information we had about her illness was only what had been communicated to us since she was a baby. It was discovered that she had blood clots in her legs, but the doctors attributed them to the hip surgery she had, since she was bedridden for a while. She began taking medication for her blood. She was examined by an ophthalmologist, and he determined that her lens had dislocated, and told us that it could have been due to a fall. Enelcita was also referred to a psychiatrist, who prescribed medication to keep her calm since her behavior varied from angry to calm. The medications

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didn't work very well and made her sleep a lot during the day. At night she didn't sleep well; she would sleep for a few minutes and her energy would return. Music became an ally for us as she would calm down for a while. For us as a family, it was very difficult. We took turns caring for Enelcita; our mother took care of her more at night and I did during the day.

It was around this time that they discovered she had gallstones, which we didn't know about since she doesn't communicate with us verbally. We always had to guess if she was in pain since she didn't have a fever and she was otherwise healthy. They stopped her blood medications because she no longer had clots in her legs.

At the end of 2019, we took Enelcita to the emergency room. We didn't know why, but she wasn't herself. It was there that they did all the tests and her blood pressure wasn't normal. When the doctors visited her and spoke to me, but I really thought they said that because of her illness, and that despite everything, she was well behaved, and it wasn't until they told us what they had found in the tests they had done that I really understood. The tests identified circulation problems in the veins that carried blood to the legs, a clot in the main vein of the aorta, and osteoporosis. Even so, we didn't know it was homocystinuria.

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It wasn't until early 2020 that Enelcita had a follow-up appointment and we met Dr. Hussan Alkaissi. He was doing his residency at Downstate Hospital and he examined her and listened to us. When he finished the examination, he mentioned homocystinuria to me and said that he had seen it before but he said, "I'm not telling you that's what she has; but I'm going to do some blood tests." When the results came back, he personally called me and confirmed that my sister did have homocystinuria. He gave me information and told me about some informational videos on YouTube. He did his best to find a genetics doctor to see her since she didn't have many options in New York. Then he found Dr. Melissa P. Wasserstein of pediatric genetics who agreed to see her and that's when we found out her homocysteine levels were at 550 if I remember correctly.

I remember very well when the doctor explained the condition to me. I cried, feeling so guilty because I fed my sister a lot of protein. I saw her as so skinny and thought she needed to gain muscle mass. Not knowing what she really had, she ate everything; she didn't follow any special diet.

From there, I searched for videos to learn more about HCU and started following the HCU Network America page. One day I attended a Zoom and heard people sharing their stories. I began to hear the stories of people with the condition and what their symptoms were like. It was there that I realized that the symptoms that Enelcita had as a child were the same, just with the wrong diagnosis. Thanks to the testimonies and a question I asked when I entered that Zoom, they helped me a lot to understand my sister much better.

“I began to hear the stories of people with the condition and what their symptoms were like. It was there that I realized that the symptoms that Enelcita had as a child were the same, just with the wrong diagnosis.”

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My mother began to better understand why Enelcita did not sleep well and the reasons for her behavior. Even though we are 4 siblings, Enelcita is the only one in the family with this condition.

Enelcita's diagnosis changed the family completely because we now had to follow a diet plan and administer medical formula, Betaine, and the rest of the medicines that she needed. My daughter and I took control of her feeding and her medications since we spoke English and could understand the doctors. My mother got the information from us since she did not speak English. It was a great relief to know that thanks to the diet and the medications, we began to see changes in Enelcita over time. Her homocysteine levels started going down and today they are at 90. Her development has been great, and she has resumed learning. Now she pays much more attention, like a growing child. She dances, sings, and likes to paint. She asks for what she wants to watch on TV and is saying words in English, which that has surprised us a lot. She does not speak in complete sentences, but we understand her.

In 2021 my mother passed away and now Enelcita is my full responsibility. But thanks to my daughter's help, we have made a lot of progress with Enelcita, even though she was not diagnosed at an early age. Now we are enjoying the process, and it makes our lives happy. Since Enelcita was born, her life began with drama. She was born in the car and did not wait to get to the hospital. From birth she shined, and she is our miracle. That is why I want to share her story. I don't know how she survived so many years without a diagnosis, how her body endured so much, and how new veins developed to bring blood to her legs. As the doctors called her "an avenger", for us she is a miracle.

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At times in the past, we experienced discrimination from some doctors, but we always kept trying new ones. Thank God the doctor showed up who was not afraid of her and who looked at her with respect. He recognized my sister's symptoms and thanks to him, we were finally able to get the correct diagnosis after so many years. Now he is finished with his residency and has left Enelcita in the care of a different doctor. Even though he is not there, the new doctor, Dr. Shiang Lin, shares Enelcita's updates with him.

If I can help others, it would be through my sister's story. If you don't have a diagnosis, don't stop seeking other opinions; the right doctor will come with the right answers. Enelcita is our hero, our miracle, and if she is still with us, it is for a purpose. My advice to people who have a family member with HCU is to be patient. Life is not always easy, but the outcome is worth it.

Click here to read Enelcita's and other patient stories on our webiste!

FOOTBALL SQUARES FUNDRAISER

FEBRUARY 9 6:30 PM ET

$10 PER SQUARE 4 CHANCES TO WIN!

$100 PAYOUTS 1ST-3RD QUARTER
$200 PAYOUT AT END OF THE GAME

TO PARTICIPATE:

1) VENMO: @TOM-HAWKINS-1
*LAST 4 DIGITS OF PHONE #: 1300

2) EMAIL TOM AFTER TO RECEIVE ACCESS TO BOARD!
TMMYHWK09@GMAIL.COM

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

PANDA EXPRESS CHINESE KITCHEN®

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HCU NETWORK AMERICA

When?

Wednesday, January 29, 2024

Place your online order for pickup or delivery on Wednesday, January 29!

Where?

Panda Express locations nationwide

www.pandaexpress.com

How?

Online orders only

Apply code **9000232** in the fundraiser code box during online checkout at www.pandaexpress.com or via App

28% of sales will be donated to HCU Network America!

LOWER-PROTEIN MEAL OPTIONS

MEAL IDEA #1:

- 1 Veggie Spring Roll = ~2 g
- Mixed Veggies (1/2 portion) + Sweet & Sour sauce = ~2 g
- 1 Fortune Cookie = 1 g

= ~ 5 g protein

MEAL IDEA #2:

- 1 Veggie Spring Roll = ~2 g
- Mixed Veggies (1/2 portion) + Sweet & Sour sauce = ~2 g
- White Steamed Rice (1/2 portion) = ~3.5 g
- 1 Fortune Cookie = 1 g

= ~ 8.5 g protein

MEAL IDEA #3:

- 2 Veggie Spring Rolls = ~4 g
- Mixed Veggies (1/2 portion) + Sweet & Sour sauce = ~2 g
- White Steamed Rice (1/2 portion) = ~3.5 g
- 1 Fortune Cookie = 1 g

= ~ 10.5 g protein

*Esitmations are approximate. For full nutritional info, visit https://www.pandaexpress.com/

Help us maximize the impact!

CLICK HERE

To visit HCU Network America + Panda Express fundraising page & share on your social media channels!

28% of sales will be donated to HCU Network America!

See previous page for details.

RARE DISEASE DAY IS COMING

Mark your calendars...

...for February 28, 2025!

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!

RARE DISEASE WEEK ON CAPITOL HILL

Registration opens January 6, 2025!

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!

Rare Disease Day is coming...

Grab your gear!

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

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

HC&U Podcast

Meet your hosts!

Welcome to the HC&U Podcast!

We are Ben and Lindsey, your hosts. We are so excited to host this podcast as extra resource for the Homocystinuria community. We hope you like our content!

You can read Ben's patient story here.

The latest episode

Ben welcomes Laurie Bonucci to the table! Laurie is the mom of Joe, an adult living with classical HCU, who was diagnosed in childhood. Laurie & Ben will chat about the impacts that Joe's delayed diagnosis had on his growth & development and how they worked to overcome these issues.

To Listen:

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

LISTEN ON Spotify | LISTEN ON iHeartRADIO | Listen on Apple Podcasts | Listen on amazon music

HOMOCYSTINURIAS DATA COLLECTION PROGRAM

POWERED BY RARE X

Growth, Developmental Issues, and Regression Surveys

from our September 2024 data pull

- 45% of those participating (32) in the Growth survey reported issues
  - 19% (11) reported tall stature
  - 9% (5) reported general growth
  - 7% (4) reported undergrowth

- 48% of those participating (32) in the Developmental survey reported issues
- 23% of those participating in the Loss of Skills/ Regression survey reported issues

Complete the Survey
https://rare-x.org/homocystinuria/

CANPKU+ CANPKU+

HCU NETWORK AMERICA

HCU Network Australia

POWERED BY RARE X

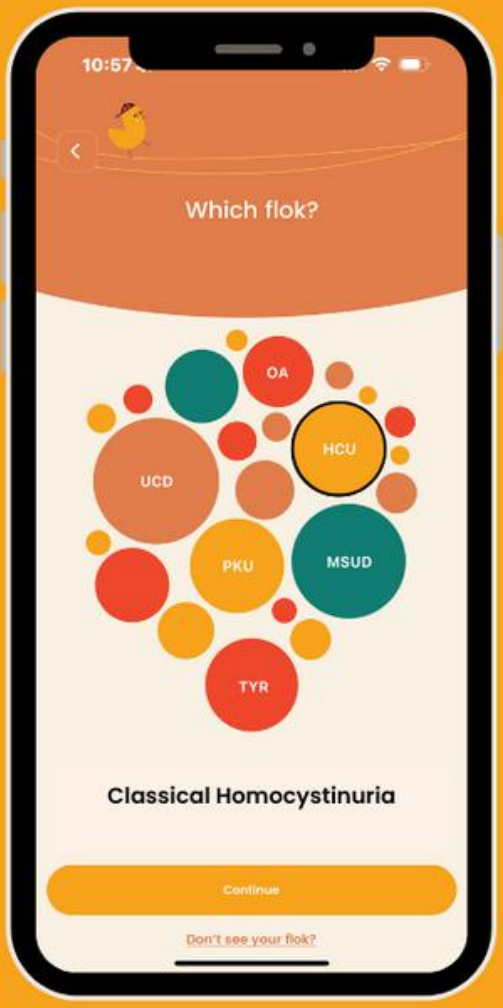
Start using the flok app today!

Welcome to the next generation of metabolic care for the CLASSICAL HCU COMMUNITY

The flok app is now in Open Beta in the United States.

Download at flok.org/app

flok



Big news!

The flok app is now in Open Beta, available for download to everyone in the U.S.!

Jump in and start exploring - the app helps you manage your diet and build a full picture of your metabolic health - including symptoms & moods, activity, medications, and lab tests.

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.

Now Enrolling

Acappella

Sponsor: Travere 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.

150

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@travere.com

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

www.hcuconnection.com.

MA-PE-24-0008. Version 2.0; en-US; March 2024

TRAVERE THERAPEUTICS

Sign our NBS Screening Petition!

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

Click here to sign the petition!

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

HOPE CONNECTS US

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