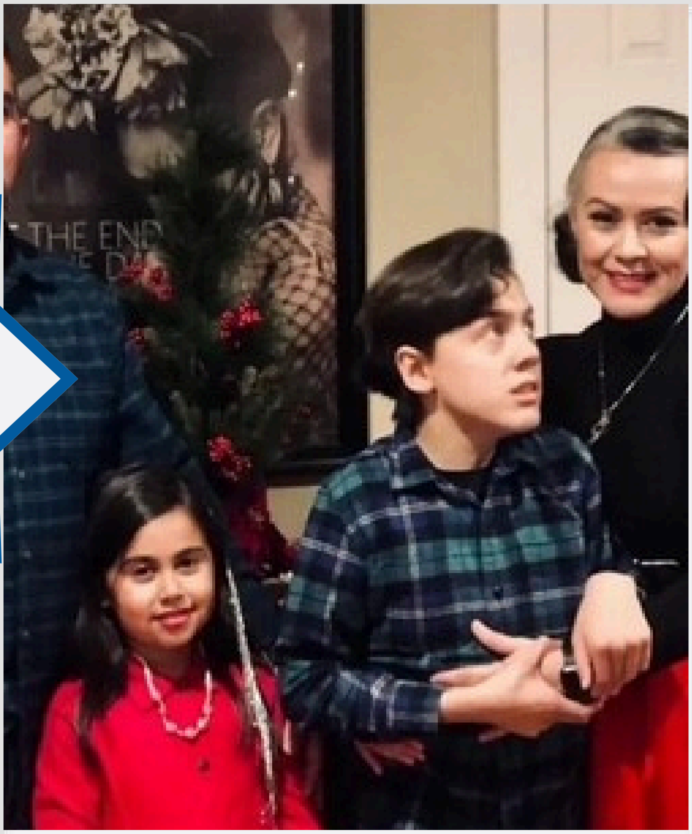
Hope Connects Us Around the World

THE HCU HERALD

Featuring...

World
Homocystinurias
Awareness
Day
Edition!!



HCU Hero

Luciano From California

May 2026

HCU NETWORK AMERICA

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

HCU HERO:
LUCIANO FROM CALIFORNIA

Luciano Ciro, came to this world on the morning of October 24th, 2009. I had a normal delivery, with no complications. The first four weeks, as I was trying to heal from the birth and bond with him, including breast feeding, his first seizure struck. At the time I didn't know how someone looked while having a seizure, let alone a newborn. I thought he was choking with the milk while breastfeeding, and was trying to gasp for air. The next day as we were visiting my brother and his wife, he had another. Thinking it was unusual and serious, my sister-in-law advised taking him to the ER. As soon as we arrived and tried to check him in, the seizures became more frequent.

That prompted the nurse to take him in quickly so the doctor could intervene. This is the moment that the ER doctor explained that what he was having were seizures, and told us that he was going to be transferred to the Children's Hospital. As all mothers know, I felt as if the rug that I was standing on was removed abruptly. He was admitted and for the first few weeks many tests were done. It was painful, not being able to continue feeding or even hold him because his condition was getting worse; failing to thrive, unable to keep body temperature stable. At that point, I was fearing the worst because the doctors couldn't figure out what was wrong with him.

Many tests were performed, including MRI scans and EEGs, to see how his brain was functioning. The neurologist found out that he had brain atrophy.

HCU HERO:
LUCIANO FROM CALIFORNIA

At that point we had a meeting with the specialist and hospital doctors so they could explain what they had found out. After 33 days in hospital, the genetic specialist had concluded after tests that what he had was Homocystinuria. The neurologist then went on to explain that due to the amount of seizures he had, his brain had suffered greatly and that Luciano would suffer from mental retardation.

That was the only thing that stuck in our heads; everything that was said before and after was a blur. I felt as if I had lost my baby and was mourning what I thought he would be like before his diagnosis. Doctors went on to say that he wanted more genetic testing to see what kind of Homocystinuria he had. We wouldn't know the results until months after Luciano had been discharged from the hospital.

We spent Thanksgiving in the hospital and on Christmas Eve we were able to go home. My life had changed. I had a newborn that would have many challenges, specifically intellectual ones. I was devastated! I was a new mother and felt isolated. I was told that very few people in the world had Luciano's condition. I tried searching the internet for anyone that was going through the same thing, but I couldn't find anything. Between therapies, doctor's appointments, and early intervention, I fell into a depression that lasted until I realized that I needed to embrace and accept my child. Throughout Luciano's first two and a half years he made progress. He started crawling at 12 months and almost 3 when he started walking with support; thanks to his physical therapist that he still sees.

HCU HERO: LUCIANO FROM CALIFORNIA

Luciano has beat all odds against him. He's resilient. He's a happy young man. Although he doesn't speak, he is very vocal and makes sure he gets his needs noticed.

Luciano suffers from all the symptoms that CblG people suffer from, including macular degeneration. Specialists have no solution than to monitor every 6-12 months. Luciano did develop serious skeletal issues, scoliosis and feet problems. Surgery was the only option to help him with that. Between his first month of life and up until now, he's been in the ICU and had ER visits due to relapse from seizures. It's been hard, always fearing when or what could cause another relapse, but Luciano is a fighter and keeps on fighting, teaching us patience, strength and resilience.

I've learned to let go of what I cannot control and enjoy the little achievements that he reaches; slowly but surely. It hasn't been easy these past 16 years. His dad and I could not keep being together, and Luciano stopped having a connection with him as well. We were fortunate enough to have met my now husband, who has stepped up to the plate and become Luciano's dad. We work together and help Luciano; from changing diapers, bathing him, giving him bike rides, administering his injections and giving him his meds.

We don't know what the future holds for Luciano, who is true to the meaning of his name...light. I am sure that whatever comes, we will be there to give him all our unconditional love and care.

Hope Connects Us Around the World

World
Homocystinurias
Awareness Day
May 18th

- **Livestream Event**
- **Exclusive Raffle Baskets**

→ Follow us to receive updated on activities & events

→ Stay tuned for livestream details & raffle information

Together, hope connects us around the world.

Syntis Bio

HCU NETWORK AMERICA

Hope Connects Us Around the World

HCU

Raffle Baskets

Click Here

WRAPPED IN HOPE

BREWING HOPE FOR HCU

STRONGER THAN THE TIDE

A JOURNEY THAT GIVES BACK

SECONDS, SPELLS, & STRENGTH

CELEBRATE RARE

RARE MOMENTS OF REST

Syntis Bio

HCU NETWORK AMERICA

Hope Connects Us Around the World

World
Homocystinurias
Awareness Day

50/50 Raffle

1 Entry	$5	+
5 Entries	$20	+

Half of the total money raised is awarded as a cash prize to the winning ticket holder, with the other half going to HCU Network America.

Click Here

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HCU NETWORK AMERICA

Hope Connects Us Around the World

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

Did you know some companies match retired employees donations?

Hope Connects Us Around the World

Celebrate

World Homocystinurias Awareness Day

In Style!

CLICK HERE

APRIL EVENT RECAP: AAN CONFERENCE

HCU Network America had a booth and poster presentation at the AAN (American Academy of Neurology) conference. Northwestern student Nicolo Benito presented data from the Homocystinuria Data Collection program on neurological symptoms of adult and pediatric HCU patients. Danae and Brittany had the opportunity to connect with Dr. Changrui Xiao (an HCUNA medical advisor) and hold a local family meet up with community members in the Chicago area!

APRIL EVENT RECAP: GMDI CONFERENCE

HCU Network America exhibited and spoke at the Genetic Metabolic Dietitians International (GMDI) 2026 conference in St. Louis, Missouri, at the very end of April.

The four-day conference brings together patient organizations, medical nutrition companies, biotech companies, and dietitians who all have a vested interest in metabolic disorders. The four days featured poster sessions, keynote speakers, and breakout sessions, with ample time to network.

During the exhibit hours, we were able to network, share resources, and provide information to help bridge the support gap between patients and clinics. We were also able to lead a workshop on “They Aren’t Just Oversized Children, Supporting Adult IEM Patients Through Life Stage Transitions.” This session provided an overview of what transition is, when it should begin, and the unique obstacles to keep in mind for patients diagnosed late.

We were able to close out our time in St. Louis by meeting two local families affected by classical homocystinuria.

UPCOMING EVENTS: KENDRA SCOTT GIVE BACK

COME SIP, SHOP, AND SUPPORT

HCU NETWORK AMERICA

AT KENDRA SCOTT, GRAND BLVD
MIRAMAR BEACH, FL

DATE & TIME:
MAY 9TH 2PM-5PM

UPCOMING EVENTS:
KENDRA SCOTT GIVE BACK

Shine Bright, Do Good

JOIN US FOR A KENDRA GIVES BACK EVENT

DATE & TIME

May 9th from 2pm-5pm

DETAILS

Shop instore at Kendra Scott Grand Blvd for 20% of the proceeds of your purchase to Give Back to **HCU Network America** on **5/9**. Can't make it? Shop online at Kendrascott.com and enter code **GIVEBACK-LXKCK** at Checkout to give back **5/9-5/10.**

KENDRA SCOTT | HCU NETWORK AMERICA

Click Here

UPCOMING EVENTS:
SPRING VIRTUAL MEET-UP

Spring

VIRTUAL

Meet Up

MAY, 30TH

11:00 AM EST

REGISTER HERE >

2026 HCU HEROES

RACE FOR RESEARCH

Cystadane; the last FDA approved drug for the homocystinurias was approved in 1996. That was 30 years ago!

Currently 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 10 research grants.

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

Race registration starts June 1!

Pricing

- Register by July 19 to receive a medal
- Register by August 24 to receive a shirt
- Early bird pricing - $30
- After July 13 - $35

RECORDATI RARE DISEASES
Focused on the Few®

HCU Network America is a registered 501c(3) foundation, registration number 81-3646006. All donations are tax deductable.

UPCOMING EVENTS

WE WILL BE AT

SIMD 2026

POSTER: "PATIENT REPORTED CLINICAL FEATURES IN INDIVIDUALS WITH HOMOCYSTINURIA: A COMPARISON OF NEWBORN SCREENING RESULTS"

BOOTH: 10

POSTER SESSION AND RECEPTION ON MONDAY, MAY 18TH FROM 7-10 PM

SOCIETY FOR INHERITED
SIMD
METABOLIC DISORDERS

HCU
NETWORK
AMERICA

HCU Network America is pleased to announce they are

Now Accepting Request for Proposals

- Seeking novel proposals for treatment and prevention of clinical manifestations for **Classic Homocystinuria** and/or **Methionine Synthase deficiency** by exploring novel mechanisms to obtain proof of concept to enable progression to clinical trials

Application Deadline: May 31, 2026

2026 HCU Network America Grant Application - Fill out form

Givinostat rescues folding of cystathionine beta-sythase and ameliorates murine homocystinuria

Maria, Petrosino, Karim Zuhra, Ela Mijatovic, Thilo Magnus PHilipp, Olivier Bremer, Kelly Ascencao, Csabo Szabo, Tomas Majtan

This research looked at a potential new treatment for classical homocystinuria, a rare genetic condition caused by problems with an enzyme called cystathionine beta-synthase (CBS). When CBS doesn't work properly, harmful levels of homocysteine build up in the body.

What problem were researchers trying to solve?

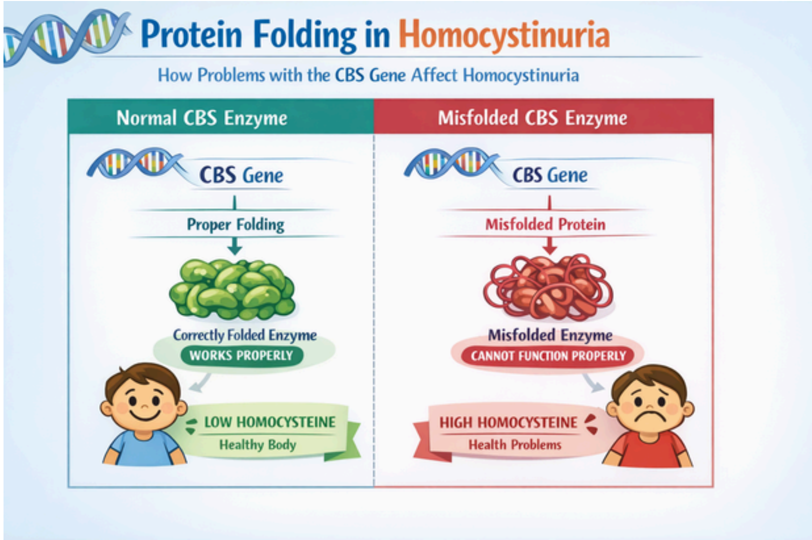
In many patients, the CBS enzyme is made incorrectly and doesn't fold into the right shape. When this happens, the body can't use it effectively—even if some of the enzyme is still there. The goal of this study was to see if a drug can be found that could help the enzyme fold correctly and start working again.

What did the researchers do?

They developed a new test enabling them to screen many drugs for their efficacy to rescue incorrectly folded CBS. They discovered a drug called givinostat, which is already being used to treat for other conditions. They tested it in cells and in mice with homocystinuria.

They looked at whether the drug could:

- Improve CBS enzyme function
- Lower homocysteine levels
- Improve health outcomes



What did they find?

- A new test allowing them to screen drugs for their efficacy to correct CBS folding.
- Improved enzyme folding: givinostat helped the CBS protein fold into a more normal shape
- Better enzyme activity: This allowed the enzyme to work more effectively
- Lower homocysteine levels: Treated mice had reduced levels of harmful homocysteine
- Improved disease features in mice: Some symptoms of homocystinuria were improved

What does this mean?

This study suggests that givinostat may act as a “protein rescue” or chaperone therapy—helping the body fix a faulty enzyme rather than replacing it.

This is important because:

- It could offer a new treatment approach for patients who don’t respond well to current therapies
- It targets the root cause of the disease in some patients

Important things to know!

- **This research was done in mice, not people**
- **It is still early-stage research**
- **More studies (including clinical trials) are needed to know if it is safe and effective in humans**

Takeaway for families

This study offers hope for future treatments that may improve how the body handles homocystinuria by helping the CBS enzyme work better. While it’s not yet available as a treatment, it represents an exciting step toward more targeted therapies.

**This study was funding through HCU Network America’s Global Grants Program

Harmony

Now Enrolling

Company: TravereTherapeutics

Total time in study: About 9 months

Goal: To learn more about classical homocystinuria (HCU), and how safe and effective pegtibatinase may be in patients with HCU

Phase: This is a Phase 3 study that follows a Phase 1/Phase 2 study called COMPOSE

Who can take part?

- √ People with a diagnosis of HCU
- √ People 12 to 65 years of age

Other factors and restrictions also apply

What is the study drug?

- √ Participants will receive pegtibatinase or placebo (an inactive medication)

Why participate in the study?

- √ You may helpotherswith HCU
- √ You will help contribute to new research and further understanding of HCU
- √ You will receive additional monitoring and testing

Study locations include: United States, Europe, the Middle East, Asia Pacific, and South America.

70

About70people with HCU will take part in the study.

Payment for time and travel may be available to participants and their caregivers.

Periods during the study

SCREENING PERIOD	PRE-TREATMENT PERIOD	STUDY TREATMENT PERIOD	FOLLOW-UP PERIOD
To check if the right study is for you	To understand your current diet and how to keep it stable during the study	To receive pegtibatinase or the placebo	To check your health after the Study Treatment Period

After completing the **HARMONY Study**, it may be possible for participants to join a long-term follow-on study and continue or start receiving pegtibatinase.

To find out more, please contact:
MedInfo@travere.com

For more information, please scan the QR code or visit:
www.hcuconnection.com

TRAVERE THERAPEUTICS

MA-PE-25-0038. V2.0_NOV2025.EN_US

WELCOME OUR NEW
Medical Advisor

Click Here

ELAINA JURECKI

Elaina Jurecki is the Director of Research Development for the National PKU Alliance and has served as a medical advisor for organizations like the Organic Acidemia Association. She consults for biotech firms on treatments for metabolic genetic disorders and previously worked 15 years at BioMarin Pharmaceutical as Executive Director of Scientific Communications. With about 20 years as Regional Metabolic Nutrition Coordinator at Kaiser Medical Centers, she has coauthored numerous publications, served as a journal reviewer, and contributed to book chapters on genetic metabolic disorders. Jurecki is a founding member of the Genetic Metabolic Dietitians International (GMDI), co-chairs various guideline development committees, and has presented extensively at scientific meetings on topics related to genetic disorders.

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

HCU FAMILY
Fun Day

On **Sunday, July 12,** to wrap up our biennial **HCU Network America 2026 conference,** we are hosting a fun way for our community to bond, laugh, and spend time together away from the hotel!

Join us for **activities and lunch** at Franklin Square from **10 am to 2 pm.**
We will be providing a continuous shuttle to and from the hotel for anyone who needs to return for departure.

If you would like to join us for this unforgettable experience, **please plan your travel accordingly.**

Carousel | Mini Golf | Yard Games

REGISTER NOW

It's A FAMILY AFFAIR

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
- Community fun day

Patient & Family Conference

REGISTER NOW

HCU NETWORK AMERICA

Classical HCU | Cobalamin Disorders | Severe MTHFR

10 REASONS
WHY YOU SHOULD ATTEND OUR
2026 HCU NETWORK AMERICA CONFERENCE

July 10-12, 2026
Philadelphia, PA

Join patients, families, caregivers, and professionals from across the country for a weekend of connection, education, and community.

1. **CONNECT WITH YOUR COMMUNITY**
   Meet others who truly understand your journey.
2. **LEARN FROM EXPERTS**
   Gain insights from leading clinicians and researchers.
3. **STAY UP TO DATE**
   Hear the latest on HCU research, treatments, and advances.
4. **HEAR INSPIRING STORIES**
   Be moved and motivated by real patient and family experiences.
5. **BUILD LASTING CONNECTIONS**
   Create friendships and networks that last well beyond the conference.
6. **GET YOUR QUESTIONS ANSWERED**
   Participate in panels and Q&A sessions designed for you.
7. **RESOURCES YOU CAN USE**
   Discover tools, programs, and support to help you thrive.
8. **FEEL SEEN, HEARD & SUPPORTED**
   You are not alone—this community has your back.
9. **BE PART OF SOMETHING BIGGER**
   Your voice and your story help drive change.
10. **LEAVE EMPOWERED**
    Walk away with hope, knowledge, and a renewed sense of purpose.

Patient & Family Conference ♡

HCU NETWORK AMERICA

REGISTER NOW

Spots are limited. Don't miss this incredible opportunity to connect, learn, and make a difference.

IN CASE YOU MISSED IT...

The HC&U Podcast is back!!!

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

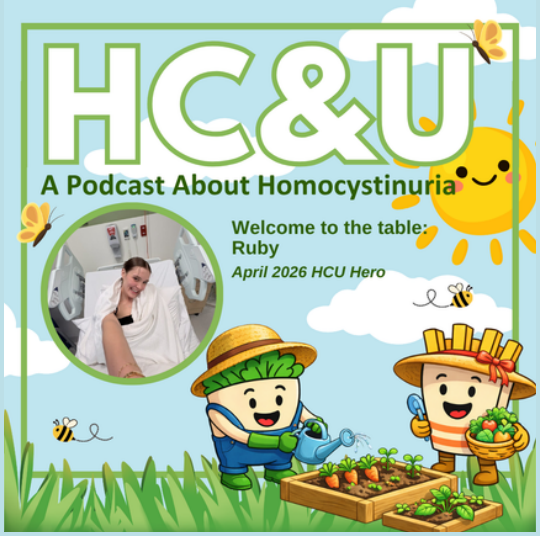
Meet your hosts!

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

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

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

The latest episode



Ben welcomes Ruby to the table! In this episode, Ben sits down with Ruby to talk about her recent experience participating in a clinical trial. Ruby shares what motivated her to enroll, what the process was really like day-to-day, and how it felt to contribute to research that could impact the rare disease community.

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

POWERED BY RAREX

The Homocystinurias affect multiple systems of the body!

The most common areas of the body affected are:

- The Central Nervous System (brain)
- Ocular (eyes)
- The Cardiovascular System (heart)
- The Skeletal System (bones)

But, many patients experience symptoms outside of these areas!

Help us to understand more:

Take the Health & Development Survey:
https://rare-x.org/homocystinuria/

CANPCU+ CANPKU+ HCU

HCU NETWORK AMERICA

HCU Network Australia

Registration Open for flok Family Camps & Adult Retreat!

Registration is now open for our 2 terrific family camps for those with inherited disorders of protein metabolism, their family members, and friends. Learn more and register at https://flok.org/camp/.

- **Family Camp West** in Antelope, Oregon, June 4-7, 2026
- **Family Camp East** on Lake Winnipesaukee in NH, September 17-20, 2026

Thanks to support from BioMarin Pharmaceutical Inc. flok's retreat and camp are **FREE** for those with Classical HCU, MSUD, an Organic Acidemia, PKU, Tyrosinemia or a Urea Cycle Disorder. Flexible pricing available for family members and friends.

Questions? Contact camp@flok.org.

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)

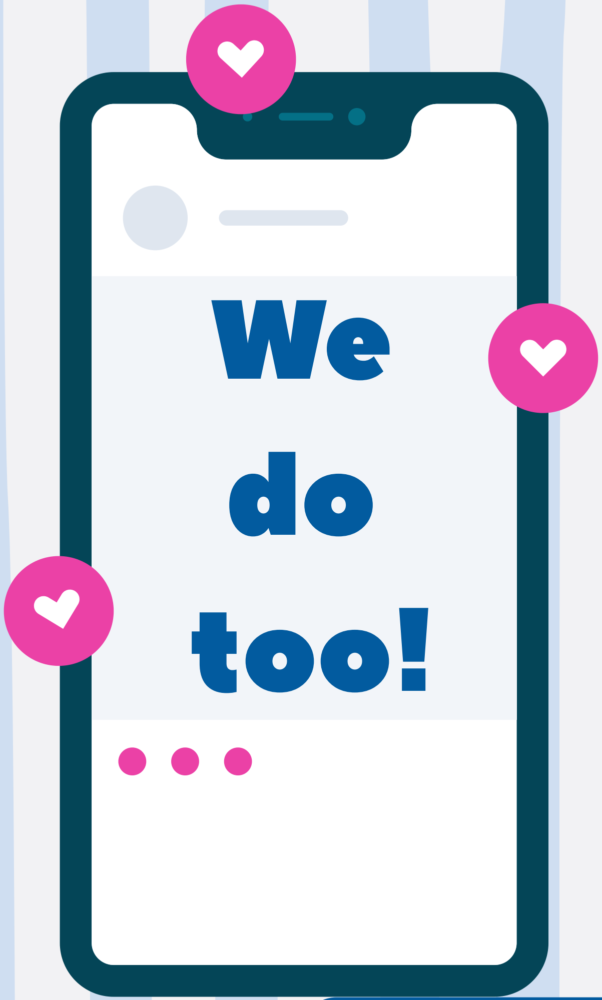
How?

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

Click Here

Follow Us!

Do you have social media??



We do too!

Follow us

@hcu_network_america

HCU Network America

@HCUAmerica